Needs Analysis of Genetics and Genomics in Communication Sciences and Disorders:

Evidence for Change

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ABSTRACT

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Purpose: Signaled by the completion of the Human Genome Project in 2003, rapid and escalating discovery in genome science has initiated a paradigm shift in education training and healthcare practices. This shift has required healthcare educators and professionals to possess a level of genetic and genomic literacy and competency. The current study was designed to survey the current state of the perceptions of genetics and genomics in educational and clinical practices within the field speech-language pathology.

Method: Seventy-five program directors of degree programs and 265 speech-language pathologists participated in two web-based surveys.

Results: Program directors and speech-language pathologist reported to be aware of recent genetic and genomic advancements in speech-language pathology. Ninety-six percent of program directors expected graduated students to demonstrate competency in genetic and genomic related clinical services. Thirty-six percent of program directors reported graduated students were prepared to understand genetics. Seventy-three percent of speech-language pathology programs offered genetic content in their curricula.

In comparison, eighty-three percent of speech-language pathologists reported performing genetic related services within their clinical practices. Less than half of respondents reported confidence in performing clinical services. Speech-language pathologists reported minimal to no knowledge of at least 85% of genetic or genomic principles related to speech-language

pathology. Sixty-three percent of speech-language pathologists reported their degree-training program had not prepared them to understand genomics in speech-language pathology. Results of a needs index revealed discrepancies between perceptions of speech-language pathologist's performed clinical services and program director's expected competencies, and between level of perceived preparedness and perceived knowledge. Thematic analysis across perceptions, course content, expected competencies, clinical services, and areas of knowledge reflected principles of Mendelian inheritance and single gene disorders. This "medical genetics" perspective is one typically used prior to the completion of Human Genome Project in 2003.

Conclusion: The results of this investigative study suggest the field of communication sciences and its disorders is not keeping pace with the demands of new advancements in genetics and genomics. Several discrepancies may contribute to misconceptions and misinformation surrounding genetics and genomic in speech-language pathology. This study provides a foundation for discussion of curriculum reform at the graduate level and policy changes in standard practices of speech-language pathologists at the national level.

TABLE OF CONTENTS

LIST OF TABLES

LIST OF FIGURES

ACKNOWLEDGMENTS

The printed pages of this dissertation hold far more than the culmination of years of study, research, and writing. These pages reflect the pursuit of a journey taken in the face of unusual adversity. That journey now ends with this dissertation and the honor of being bestowed a Doctorate of Philosophy.

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Chapter I

INTRODUCTION

Statement of the Purpose

The purpose of this research is to characterize the current state of genetic and genomic education in the field of communication sciences and its disorders. This study seeks to provide evidence of the current state of genomic education in degree programs and work practices of professional speech-language pathologists. The evidence will provide an understanding of areas in need of a change, and provide a foundation for further research in genomic education in communication sciences and its disorders. The findings will contribute to a discussion of curriculum reform at the graduate level, and national policy changes in the standard practices to include genetic and genomic literacy and competency.

Specific Aims

The specific aims of this study were to:

(1) Characterize the current state of genetic and genomic education in the educational training of speech-language pathologists as perceived by program directors:
   (a) Characterize program director's perceptions of genetics and genomics
   (b) Identify and characterize genetic and genomic content in curricula of degree programs

(c) Identify and characterize qualitative and thematic concepts

(2) Characterize the current state of genetic and genomic education, knowledge, and clinical practices as perceived by the professional speech-language pathologist:

(a) Characterize the professional development education with regard to genetic and genomics

(b) Characterize perceptions of genetics and genomics

(c) Characterize the perceived knowledge in genetic and genomics in speech-language pathology

(d) Identify the clinical services within scope of practice as they relate to genetics and genomics

(e) Characterize the confidence levels of speech language pathologists as they relate to genetic and genomic related clinical practices

(f) Identify and characterize qualitative and thematic concepts

Significance and Rationale

The completion of the Human Genome Project in 2003 has led to rapid and escalating discovery in genome science. What has resulted is a paradigm shift in healthcare and communication sciences and its disorders (Robin, 2008, p. vii). Every health condition, disease, disorder, and behavioral trait is now thought to have a genomic basis; that is, a complex interplay of genetic and environmental factors (Collins, 2010a; Collins, Green, Guttmacher, & Guyer, 2003). Consequently, health care professionals, including speech-language pathologists, must begin to view communication and its disorders through a genomic lens (Collins, 2010b; Robins, 2008).

As a profession, speech-language pathology has been lacking in developing a workforce prepared to deliver genomic-based care knowledge (Chermak & Wagner-

Blitz, 1993; Christianson, McWalter, & Warren, 2005; Robins, 2008). There is presently little published information about the level of genetic and genomic knowledge among professional speech- language pathologists. The same lack of published information is present in the education of speech-language pathology students. There is little known of the genetic and genomic education in speech-language pathology programs.

The training programs in speech-language pathology represent the entry point of the continuum of education. Training continues with the professional development of the speech-language pathologist in clinical practice. The program directors of speech-language pathology training programs were an identified group in the study. Program directors are in the position to have an overview of the faculty and student body, in which the speech-language pathology curriculum is offered. The program director's responsibilities are to lead, guide, and support the program and the faculty in directions that may influence the curriculum content.

Students and faculty of a training program are acknowledged as integral stakeholders in the survey; however, they were not the subjects of study in the current research. Professional speech-language pathologists were targeted as they represent the end product of the training program; it is they who will be engaged in genetic and genomic related clinical services. They are mandated by national and state regulations to maintain continued professional development.

The purpose of this investigation is to survey and identify the current state of genetic and genomic education in communication sciences and its disorders. Two target groups were selected to survey: professional speech-language pathologists, and academic degree programs in speech-language pathology. The results of the surveys will identify areas of need or discrepancy in the current state of genomic education, and will provide evidence for discussions for curriculum reform to include genomics. It will also provide evidence for calls for changes of professional policies in the standards of practice of professional speech-language pathologists.

Arrival of the Genome Era

The completion of the Human Genome Project (HGP) in 2003 publicly heralded the dawn of the genomic era in health care. Advances in genetics and molecular biology developed in the context of the HGP have demonstrated a genetic or genomic component to all diseases and health conditions (Ferro, Guttmacher, & Collins, 2010). Medical genetics, itself a relatively new field of study, has given way to human genomics. Genomics is the study of how the total DNA complement of an individual or a population, in concert with all manner of environmental factors, contributes to all things human – development, behavior, health, disorders, diseases, and illness.

Whereas genetics focuses on relatively rare single gene disorders, genomic science embraces behaviors, diseases, and disorders of complex and multi-factorial origin. Simply stated, genomics takes a holistic view of genetics. Genome-based approaches are playing an increasingly important role along the health continuum in disorder/disease prevention, screening, diagnosis, management, and treatment. Genomics has become a central science in health care (Consensus Panel, 2006). As evidenced in other fields of medicine, nutrition, psychology and social sciences, the translation of research findings into clinical practice is occurring much slower than the blinding pace of knowledge acquisition. (Bankhead, Emery, Quresh, Campbell, Austoker & Watson, 2001; Dougherty, 2009; Farndon & Bennett, 2008; Iredale & Cleverly, 1998). This leads to a growing knowledge-to-practice gap in genetics and genomics, which is faced by all healthcare practitioners, including speech-language pathologists.

Speech-Language Pathology's Role

Speech-language pathologists in the United States stand at the intersection of escalating advances in genome science, and an increasingly genome literate healthcare field. The translation of genomic science into healthcare practice has required speech-language pathologists to become familiar with new terminology, concepts, skills and

technologies. In fact, genomic science provides a new lens through which speech-language pathologists can view communication sciences and its disorders. The approach of medical genetics, in which inheritance of relatively rare gene alterations directly causes specific disorders, has given way to a much broader genomic view (Dougherty, 2009). To see speech-language disorders through a genomic lens requires the educator and the clinical speech-language pathologist to consider the collective influence of multiple gene variations and the cumulative effects of all manner of environmental factors, susceptibility or risk for disorders, and response to therapies. The genomic era represents a paradigm shift to which the field of speech-language pathology must respond.

There is little doubt that the field of speech-language pathology benefits from becoming genome literate and competent. Rationales for achieving genomic literacy and competency include:

1. Genome science is being translated to clinical practice across all healthcare settings: medicine, nursing, social work, occupational therapy, nutrition, psychology, etc. Evidence-based practice requires speech-language pathology to apply research findings to patient/client care and management. Anticipating increasing application of genome science in healthcare delivery, speech-language pathologists must prepare to provide care based on awareness of genomic influences, as well as become genome literate collaborators with other healthcare providers.

2. Speech-language pathology education must prepare graduates to keep pace with changes driven by research and technology and provide both leadership and education as clinical, ethical, legal and social implications of genomic health care evolve.

3. Speech-language pathologists are compelled to engage in lifelong learning and continual professional growth in order to achieve and maintain knowledge relevant to the current scope and standards of practice. Consequently, speech-language pathologists

have a duty to update their own knowledge as the science of speech-language pathology changes.

Like any change in the health care environment, the arrival of the genome era creates uncertainty and "role ambiguity" for the professional (Jenkins, 2000). Speech-language pathologists in all settings, including clinical practitioners, students, educators, researchers, administrators, and policymakers, must recognize the relevance of genomic knowledge and then work to achieve genomic literacy and competency. Speech-language pathology educators in the degree programs and those providing professional development face an enormous task to deliver genetic and genomic knowledge to the clinical workforce.

Genetic/Genomic Knowledge among Speech-Language Pathologists

In many ways, speech-language pathologists are well prepared for the activities that comprise genomic healthcare, having specific training in history taking, multidimensional assessment, and effective communication of complex information. However, research, although scant, has consistently documented a lack of genetic knowledge among practicing speech-language pathologists (Chermak, & Wagner-Blitz, 1993; Christianson et al., 2005; Lapham, Kozma, Weiss, Benkendorf, & Wilson, 2000). Therefore, as genomic science is increasingly translated into healthcare practice, the gap between what speech-language pathologists know and what they need to know may be growing wider.

Genetic/Genomic Knowledge of Speech-Language Pathology Students

Students are expected to come to degree programs in speech-language pathology with a fairly broad understanding of genetics. In the United States, National Science Education Standards (NSES) established in 1996 recommended specific genetics concepts to be included in science education in grade and high school (Center for Science, Mathematics, and Engineering Education, 1996). Although the NSES are

thought to be influential, they are only guidelines, and individual states set their own science learning standards. In addition to the genetic and genomic content students receive prior to college, natural science courses required by many degree programs as prerequisites potentially offer further opportunity for genetic and genomic education. However, no recent data are available about the level of genetic and genomic knowledge among entry-level undergraduate and/or graduate students.

It is reasonable to think the level of genetic knowledge at the entry level of a graduate degree in speech-language pathology students may be similar to that of other undergraduate college students. However, a review of the literature is does not reveal the genetics knowledge among the general college population.

The Core Competencies

In 2004, a group of organizations involved in healthcare established a minimum basis for preparing the speech-language pathology workforce for the genome era. The National Coalition for Health Professional Education in Genetics (NCHPEG, 2004, 2007) published a set of 52 competencies (Core Competencies in Genetics Essential for all Speech-Language Pathology – referred to as the *Core Competencies),* which translated genetic knowledge, skills and attitudes into speech-language pathology activities that collectively constitute genetic- and genomic-based knowledge. The *Core Competencies* have been endorsed by the professional organization, the American Speech-Language Hearing Association (ASHA). The *Core Competencies* were developed to provide the necessary benchmark for establishing a level of genetic and genomic knowledge of speech- language pathologists. In other words, the *Core Competencies* reflect the current critical knowledge that constitutes genetic and genomic literacy and competency in speech-language pathology.

Measuring Genetic/Genomic Knowledge

Three previous investigations of perceived genetic knowledge among practicing speech- language pathologists have decidedly a genetic focus and have measured perceived knowledge of basic science of genetic principles (Chermak & Wagner-Blitz, 1993; Christianson et al., 2005; Lapham et al., 2000). To date, no published studies have systematically assessed the curricula of training programs in speech-language pathology with regards to genetic and genomic content, nor has genetic and genomic knowledge and clinical services among speech-language pathologists been assessed using genetic and genomic principles and the *Core Competencies* as the benchmark. Furthermore, no validated instrument is currently available to measure knowledge underlying the essential speech-language pathology genetic and genomic competencies.

A Summary of What is Not Known

The potential for genomic advancements in the field of communication sciences to have greater role in the clinical practices of speech-language pathologists is only as salient as its ability to translate the advancements into a genome literate and competent field of speech- language pathologists. However, there are several challenges in making this translation occur. There exists little knowledge about the current state of genomic literacy and competency in the field of speech-language pathology. The following reflects what is unknown:

1. What is the genetic and genomic knowledge (literacy and competency) of professional speech-language pathologist?
2. How do we measure genetic and genomic knowledge?
3. Require a consensus of *what* speech-language pathologists need to know in regards to genetics and genomics?
4. Are genetic and genomic related clinical practices in use by the clinician?
5. Are speech-language pathology degree programs offering genetic and genomic content in their curricula?

6. What is the consensus of what and how genetics and genomics should be taught in a training program?
7. What is the genetic and genomic knowledge of the student entering in an undergraduate and masters level degree program?
8. What is the genetic and genomic knowledge of the graduating student of an undergraduate and masters level degree program?
9. What are the perceptions, attitudes and opinions of program directors, faculty, and students?
10. What are the perceptions, attitudes and opinions of professional speech-language pathologists?
11. What are the perceptions, attitudes and opinions of professional association policymakers?
12. What are the perceptions, attitudes and opinions of higher education policymakers?
13. What standards exist regarding genomic curricula requirements?
14. What is the literacy rate of communication and its disorders of geneticists, pediatricians, and otolaryngologists, of the general public?

A Summary of What This Study Will Contribute

Evidence needs to be gathered as to what is perceived to be known and not known in regards to the education of two identified stakeholders; degree programs educating future speech-language pathologists, and the professionals who are providing clinical care to individuals with genomic-based communication disorders. This study will provide preliminary evidence for understanding the current state of genomic education as per the perceptions of program directors. The investigation will also identify the genomic education and perceived knowledge, thereby establishing a consensus of *how* speech-language pathologists are being educated, and *what* is being taught during the continuum

of professional education. As well, it will provide evidence of *what* the field of speech-language pathology perceives it knows, and what it needs to know.

The findings of this research will assist in the important next step of development of a genomic literate and competent profession, providing evidence to inform national policy makers on standardizing customary genomic practices in the education and professional development of speech-language pathologists.

Chapter II

LITERATURE REVIEW

Literature across multiple disciplines informed this research and is described here. First, concepts underlying genomic speech-language pathology education are presented, in order to create a clear foundation for discussion. Attention is then turned to the changes in genetics, genomics, and communication sciences that over the last decade have led to the need for genomic education. The current state of genetics and genomics in communication sciences is then explored in order to understand the context in which needs assessment is warranted to effect change. This broad review of the literature is necessary to understand, in context, the extent of the current state of genomic knowledge in the field of speech-language pathology.

Conceptual Framework

David Ausubel, whose theory of concept and learning assimilation provides a theoretical framework for this study, defines *concepts* as “objects, events, situations or properties that possess common criterial attributes and are designated by some sign or symbol” (Ausubel, Novak, & Hanesian, 1978, p.56; Woolfolk, Perry, & Skapk, 2010). Conceptual and proficient understanding is not static. In the last decade, the concept of genetics has changed with acquisition of new knowledge borne of the Human Genome Project. During that time the terms *genomics* and recently, *epigenetics* has permeated

scientific, professional and popular writing. Unfortunately, the meanings of *genetics* and *genomics* are often conflated, even in professional literature (Ward, 2011). Efforts to resolve concept ambiguity around genetics, epigenetics, and genomics must occur early in the process of implementing genomic education. Other concepts have emerged during recent years as well, that warrant explication: *genomic taxonomy, genomic health care*, and *genomic literacy and competency*.

Genetics

Genetics is the study of individual genes and their protein products (Guttmacher & Collins, 2002). As Ward (2011) describes, genes, as functional and physical units of DNA (deoxyribonucleic acid), are sequenced and located at specific sites along chromosomes. The role of a gene is to direct the formation of one or more functional proteins in amounts adequate to support normal physiologic activities. A change in DNA sequence within a gene (known as a polymorphism or mutation) may cause the formation of a defective protein with altered function. Thus, a typical genetic condition occurs when a single gene, or both copies of gene alleles (see Glossary in Appendix Q), carries a mutation that interrupts normal gene function. This is the basis for traditional genetic conditions.

Medical genetics, the study of heritable diseases, is a relatively new discipline, born after the end of World War II on the heels of the ignominious eugenics movement (Collins, 2010a). The heritable disorders addressed by medical geneticists are fairly rare and mostly single-gene disorders of predictable or Mendelian inheritance. Traditional genetic diseases include Down syndrome, phenylketonuria, cystic fibrosis, cleft lip and palate, and Huntington disease. There are over 4000 single gene disorders identified to date (OMIM, 2011).

Epigenetics

In genetics, gene expression is the most fundamental level at which the genotype (the molecular expression of genes) gives rise to the phenotype (the physical-behavioral expression of genes and environment). The genetic code for expression stored in the DNA is “interpreted” (gene expression), and the properties of the expression give rise to the organism's phenotype.

The field of “epigenetics” identifies the instructions (mechanisms) of gene expression (changes in the chromosome) caused by mechanisms other than changes in the DNA sequences. These instructions are important for normal functionality while their malfunction may lead to ageing, cancer, diseases, and maladaptive behavioral traits (Plomin, 2003).

There are many types of instructions that effect variances in gene expression. For example, Prader-Willi syndrome (see Glossary in Appendix S) is caused by genetic and epigenetic mutated errors (deleted chromosome material) to part of chromosome 15. If proteins change certain instructions, mutated DNA received from the mother will result in a child with Angelman syndrome (see Glossary in Appendix S). If the same instructions are received from the father, the disease will result in Prader-Willi syndrome. The same deletion on the long arm of chromosome 15 occurred in both Angelman syndrome and Prader-Willi syndrome, however due to epigenetic *“imprinting”* or different male and female methylation patterns or instructions, the disease will be expressed differently, resulting in two different expressed syndromes.

Another example is offered; in times of drought, the body produces molecules to modify DNA and “*turn on or off*” through a methylation instruction and *“turn the dial for more or less”* through histone modification instruction (genes that assist in enduring difficult circumstances). In other cases, interfering molecules come from the environment. Molecules called methyl groups are present in foods, household chemicals and environmental pollutants. These can modify the structure of DNA through

methylation, turning genes on and off and affecting what gets translated into RNA and proteins, thus affecting processes of aging and susceptibility to cancer and other diseases and disorders.

A new field of epigenetics has recently developed, called behavioral epigenetics. One of the most stunning discoveries in behavioral epigenetics is the generational effects of maternal bonding. Champagne (2008, 2011) showed that rats that spent more time grooming their young resulted in those offspring braver and more resilient to stress. The infant rats actually changed their behavior due to epigenetic effects when their mother's grooming caused a particular methylation pattern in the murine pups' brain. These changes were evident in subsequent generations of offspring.

Epigenetics is promising great potential in how its study will provide a better understanding of all human behavior, including communication and its disorders. Just as genomic sequencing and the Human Genome Project revolutionized the last decade of scientific research, the next several decades may prove to be dominated by epigenetics.

Genomics

Although the term *genome* has been used for nearly a century to indicate the entire DNA sequence of an organism, *genomics* is a much newer concept. The roots of genomic medicine lie in the decoding of the human genome, accomplished in 2003. *Genomics* is the study of the total DNA complement of an individual or a population, including environmental effects on gene expression. Genomics involves not just the small fraction of DNA contained in genes, but also DNA that lies between genes and does not encode protein. Genomics addresses these and other contributors to gene expression, including interactions between genes as well as effects of environmental factors. Ward (2011) offers this analogy – if genomics is like a garden, genetics is like a single plant. If the plant isn't flowering, you could study the plant itself (genetics) or look at the

surroundings to see if it is too crowded or shady (genomics-including epigenetics) – both approaches are needed to understand how to help make your plant blossom.

Therefore, whereas a typical genetic condition is related to malfunction of a single gene, genomic conditions develop due to contributions of multiple genes and are often modulated by environmental effects. Evidence of genomic expression is accumulating for virtually all diseases, disorders, and behaviors. Common chronic disorders such as hypertension, diabetes, asthma, heart disease, and cancer have genomic causes. However, so too do autism, reading disorders, learning disorders, and speech and language disorders. That is, genetic traits (nature) in concert with multiple environmental factors (nurture) contribute to gene expression and possible alterations in health and behavior (epigenetics). All traits, all diseases, and all disorders have a relationship to genomics.

Jenkins and Calzone (2007) caution that the definitions of genetics, epigenetics, and genomics must remain works in progress since ongoing research will change the understanding of genome science. Accordingly, the translation of genetic and genomic advances to the field of speech language pathology needs to adopt appropriate reference to the *"genomics"* in speech- language pathology and not *"genetics"* in speech-language pathology.

Genomic Taxonomy

The Human Genome Project (HGP) represents a significant change in the evolution of science, and in doing so has contributed to the recognition that these advancements are changing our understanding of the concepts of the gene, genetics, the epigenome, epigenetics, the genome, and genomics. The taxonomy of what has been referred to since 1856 as "genetics" has changed and is now considered to represent three specific eras in time; the years represented in the generations from 1866 to 1990 has been referred to as the pre-genomic era; the genomic era is considered from 1990 to 2003, and the post-

genomic era, which is considered to have begun from the completion of the HGP in 2003 to the present. The post-genomic era is being considered by some to be expected to last much longer, probably extending over several generations (Collins, 2010ba).

The Pre-Genomic Era

Genetics in the pre-genomic era has been often referred to as *classical genetics* where the focus was placed on delineating the "gene", on understanding Mendelian modes of inheritance, on explaining single gene disorders, and in its practice of medical genetics--assessing, diagnosing and treating inherited diseases and disorders (Collins, 2010; Robins, 2008). In the early period of the pre-genomic era, genes and genetics as a theory of heredity functioned as an investigative tool with which geneticists could explore broader biological questions. During the 1980s and 1990s, genetics experienced a division of biologic genetic science that was centered around the practice of medical genetics (Robins, 2008). Medical genetics had traditionally focused on those conditions that were known to be due to mutations in single genes (e.g., Huntington disease, velocardiofacial syndrome), whole chromosomes (e.g., Trisomy 21 in Down syndrome), or associated with birth defects and intellectual disabilities. For these conditions, a traditional single gene genetic model applied with its accompanying processes (assessment, diagnosis, counseling, and management).

The Genome Era

In contrast, the genome era spans a shorter time frame – it comprises a 13-year effort by the government sponsored academic centers and an independent corporation called *Celera,* to determine the sequence of chemical base pairs, which make up DNA (U.S. Department of Energy, 2012). Additional efforts were placed on mapping the approximately 20,000-25,000 genes of the human genome from both a physical and functional standpoint. A working draft of the genome was announced in 2000 and a completed draft was presented in 2003. The "race" to sequence the human genome

resulted in the development of new technologies in genetic research. These developments have contributed much of the depth and wealth of genetic and genomic information now known (U.S. Department of Energy, 2012; Ward, 2011).

The Post-Genomic Era

The post-genomic era is the time period in which the completion of the HGP resulted in a dramatic increase in the amount of sequenced data publicly available, not only in the human but also including many whole genome sequences of various plants, mammals (i.e., horse, dog, cat, gorilla, etc.), and bacteria, including *treponema denticola* – the bacterium found in dental plaque causing gum disease (Wikipedia.org, 2012).

In the early period of the post-genomic era significant challenges to conventional assumptions about the relationship between genome structure and function, and between genotype and phenotype were raised, bringing about further development and understanding of both genetics and genomics. There exists a quantitative difference between the two fields (the study of multiple genes vs. one gene, which could make genetics part of genomics). Yet the practice of genomics centers on information resulting from variation on one or multiple chromosomal loci and strong interactions with environmental factors (broadly defined to include stress, nutrition, drugs, infectious agents, physical agents, and behavioral factors). In addition, there is a qualitative shift between genetics and genomics in its applications. This shift ranges from the concept of a disease or disorder in genetics, to the concept of information in genomics, a better understanding of the integration of environment (Collins et al., 2003). In many ways this dichotomy can be viewed as concrete, however, in real life applications it may be best viewed as a continuum, with no clear distinction from single gene disorders with high penetrance to genetic information obtained from multiple loci in somatic cells. With multiple genome sequences available, and the development of new technology, new fields of genomics are being developed. The advancements has demanded new

nomenclature, new etiologies of disease and disorders, redefined classification systems of diseases and disorders, new management protocols, new therapies (stem cell, genopharmacologic), new policies, and new laws (GINA, 2008). Figure 1 is a schematic of the Pre- and Post-Genomic Eras.

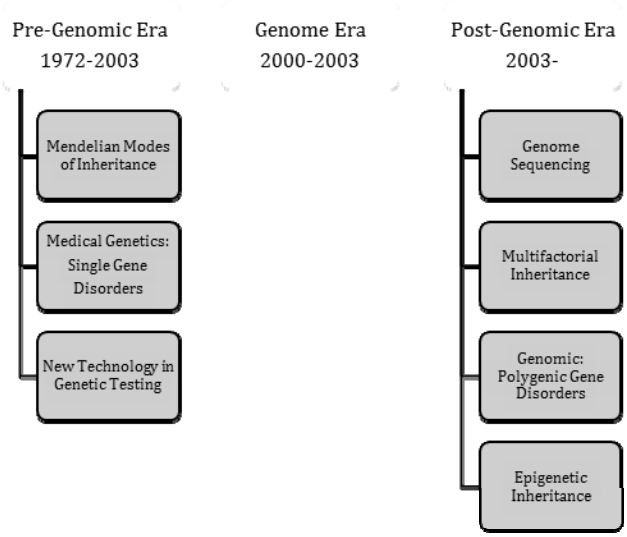

Figure 1. ***The Genomic Era Taxonomy Model***

By the time the Human Genome Project had been completed, the human genetic code became available to all in a public database (HGP, 2003). However, the HGP provided more than the human DNA sequence. An additional result was the refinement of technology necessary to mine that code for variations associated with human health and illness. It was purported that genomic knowledge would allow the prediction of specific disease risk among individuals, families and populations, encompassing not only genetic influences on health, but environmental factors as well (Collins et al., 2003).

Discussions of ‘hope’ versus ‘hype’ have become common in genome literature, as clinical applications of genome science have been far outpaced by dazzling progress in genome research (Robin, 2008; Ward, 2011). While the transformation of healthcare that was predicted upon completion of human genome sequencing may yet occur, genomic science has encountered delays in translation to practice.

Although speech-language pathology has not yet been radically transformed by genomic science, effects of genomic science have begun to permeate all aspects of care (LeBlanc, 2010). Genomic healthcare is simply care tailored to an individual based on genetic and genomic information (Green, 2010). For speech-language pathologists, genomic healthcare could incorporate genomic tools into clinical practice (however, these tools have yet to be operationalized). Genomic tools are not all high-technology molecular manipulations: On the contrary, one of the most fundamental tools, the three-generation family history (pedigree analysis) is also the simplest, requiring only pen and paper. However, genomic healthcare involves more than a set of skills. It also requires speech-language pathologists to adopt a new theoretical view of *"thinking genomically"* to see communication and its disorders through a *"genomic lens."*

Current knowledge of genomics incites an acute awareness of the interface of heredity and environment. Individual patient/client characteristics including race and ethnicity, risk for disorders, response to therapy, even genes and environmental factors acting together have an effect on behavior. Does a family share a predilection for stuttering because of common learned behaviors? Or is the person who experiences uncontrolled stuttering under the effects of reversible metabolic lysosomal changes (a genetic variation that prevents metabolism) or familial inheritance? Is the child with delayed onset of speech, one who is "lazy" and non-adherent to the appropriate speech-language model in its environment? Or might s/he have altered genes that interfere with appropriate neuronal growth and maturation? To view a patient/client through a genomic lens is to recognize and consider the interplay of nature and nurture on communication and its disorders.

Genomic Literacy and Competency

A handful of speech scientists have championed genetic research in communication sciences in recent years; Lawrence Shriberg, Barbara Lewis, Bruce Tomblin, and Mabel Rice (LeBlanc, 2010). Yet, the addition of genetic and genomic research to the field of speech- language pathology has resulted in a realization of the need to translate genetic research to the field. Genomic advancements have highlighted areas of possible deficiencies in genetic and genomic content in sectors of education and work practices. Not every speech-language pathologist needs to be a genetics expert, but there is content speech-language pathologists must know in order to provide competent services. A content outline has been proposed the *Essentials Genetic Competencies for Speech-Language Pathologists* (NCHPEG, 2007), which represents current consensus on the set of knowledge and skills required by speech-language to deliver competent care based on understanding of genetic and genomic influences. However, it is crucial to note that the *Core Competencies* are written as competencies and do not specifically delineate the fund of knowledge required for speech- language pathologists to achieve genomic proficiency (skill). In a number of domains, terms such as literacy and competency are used interchangeably to describe knowledge or proficiency. In general, literacy is more closely aligned with knowledge, while competency infers the ability to apply that knowledge. The terms are, however, sometimes conflated and warrant examination to provide conceptual clarity.

Literacy

Literacy in general is understood to be knowledge content and context specific (Ratzan & Parker, 2006). E.M. Rogers's (2003) theory-based framework distinguishes three types of increasingly complex knowledge; "awareness knowledge," which refers to knowledge about the existence of an innovation; "how-to-knowledge," which is practical knowledge concerning the proper use of an innovation; and "principles knowledge," or

integrated understanding of underlying theoretical principles of the innovation (Rogers, 2003; see Figure 2). Distinguishing these types of knowledge allows a more sophisticated understanding of the quality and quantity of genetic and genomic content relative to the field of communication sciences.

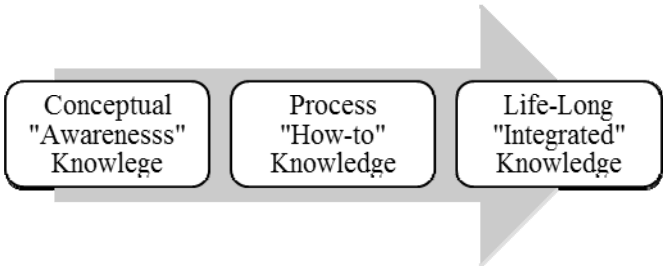

Figure 2. ***Rogers's Model of Knowledge***

Application of Rogers's knowledge framework to current knowledge concepts of genetic and genomic in communication sciences will provide valuable insights on the structure of known and unknown.

Much has been written about the body of knowledge that constitutes genetic or genomic literacy. Kaphingst (2009) considers genomic literacy in terms of health literacy, applying the definition that was used in both Healthy People 2010 and the 2004 Institute of Medicine (IOM) report on health literacy: "Health literacy is the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions" (IOM, 2004; Ratzan & Parker, 2000, p. vi). This is a functional definition, describing a level of understanding necessary to support a specific role. So defined, literacy cannot be achieved by rote learning alone but requires a level of knowledge that reaches the application level or higher in Bloom's taxonomy (Anderson & Krathwohl, 2001; Ward, 2011).

Ratzan and Parker (2000) describe health literacy as the currency needed to navigate an increasingly complex health care system. Genomic literacy for the speech-

language pathologist might be considered similarly, i.e., as the currency necessary to apply genomic principles in the context of one's personal and/or professional roles. The degree of understanding that constitutes genomic literacy varies according to the context in which it is applied. For example, Jennings (2004) suggests that the public require genetic literacy sufficient for informed consumerism, i.e., to be able to provide informed consent for a genetic test or gene-based therapy.

For the speech language pathologist, however, the literacy requirement is greater. Speech-language pathologists have a role in the delivery of genetic-related services and management of genetic and genomic information, and a responsibility to advocate for patients/clients within the healthcare and educational system. Speech-language pathologists also have a duty to provide leadership in healthcare policy decision-making, which may increasingly address genetic and genomic issues. The various activities by which speech-language pathologists provide genomic health care are not yet fully realized. For the field of communication sciences and its disorders, then, genomic literacy requires knowledge sufficient to carry out the activities that make up those competencies.

Speech-language pathologists, therefore, require conceptual knowledge that underlies the competencies, along with the specific language required to understand and articulate issues in genomic healthcare. The delivery of genomic healthcare does not require speech-language pathologists to have detailed knowledge of genetic mechanisms. It does, however, require an understanding of genetic and genomic terminology and a solid grasp of the underlying concepts of genome science for clinical practices.

Speech-language pathologists must understand that genetics also has a role in driving behaviors, since studies in behavioral genomics have linked gene polymorphisms with attention, learning, reliance, motivation, anger, risk-taking behaviors, and optimism (Plomin, Defries, Craig, & McGuffin, 2003). Genomic literacy supports clinical activities such as genomic-based assessments and testing, genomic-based therapies, and possible

pharmacogenomic and nutrigenomic applications, and a greater understanding of the role of genetics in human behavior.

Competency

In the last half-century, competency-based educational frameworks have become dominant in health professional education, largely replacing knowledge-based models (Carraccio, Wolfsthal, Englander, Ferentz, & Martin, 2002). The competency approach has evolved side-by-side with, and perhaps been driven by, the development of national educational and practice standards which delimit the set of knowledge, skills and attitudes thought to represent competence in a particular domain. The shift to competency-based education has raised important questions about what it means to be competent and how (or even if) competency can be measured.

The Centers for Disease Control, in developing competencies for public health professionals, defined competencies as "applied skills and knowledge (blended with behaviors)" that enable effective practice (Centers for Disease Control [CDC], 2001). Based on allied health education meta-analysis, (Carraccio et al., 2002) defined a competency as "a complex set of behaviors built on the components of knowledge, skills and attitudes" and defined competence as "personal ability" (p. 362). Whitcomb (2002), writing about medical residents, pointed out that competence implies the ability to provide services "in accord with practice standards established by members of the profession and in ways that conform to the expectations of society" (p. 359). He suggested that knowledge, skills, and attitudes are necessary but not sufficient to achieve competency. Also required is the ability to translate knowledge, skills and attitudes into a set of complex behaviors that result in the delivery of high-quality care.

This translation requires critical thinking, decision-making and interpersonal skills. Benner (1982) noted that competency implies not only skill application in real situations,

but also desired outcomes: “Competency ... is the ability to perform [a] task with desirable outcomes under the varied circumstances of the real world” (p. 304).

Genomic literacy is necessary but not sufficient for genomic competence. As Benner (1982) reminds us, competence develops over years, not over a semester, or a conference day. However, the development of genomic literacy might very well occur during a semester, or a year, or a curriculum.

Genetic and Genomic Advancements in Communication Sciences

The year 2010 marked the beginning of the second decade in what has been referred to as the ‘genetic informational age.’ It was the 10th anniversary of the initiation of the Human Genome Project (HGP) (Collins, 2010; van Ommen, Bakker, & den Dunnen, 1999). In 2003, a conference was held at the National Institutes of Health in Bethesda, Maryland announcing the completion of the sequencing of the human genome-The Human Genome Project (Collins, 2003). It was exactly fifty years earlier that Watson and Crick first described the structure of the DNA molecule (Watson & Crick, 1953).

The genetic advances in scientific and technological knowledge resulting from the HGP are unprecedented. They continue to yield significant findings in genes and what they do (genetics) and the collective role they play (called genomics). We now know more about genetics, genomics and human behavior than in any other time in history (Feero, Guttmacher & Collins, 2010a; McInerney, 2002).

A new direction in the relationship of genetics and communication sciences was crystallized in 2001 with the finding of a gene, called forkhead-box P2 (FOXP2) (OMIM[1] 605315). The FOXP2 gene was implicated in the disordered speech of a 3-

[1]OMIM-Online Mendelian Index of Man is an online data-base of human genes and genetic disorders.

generation British family. The KE family, as they are known, was discovered to have a particular gene mutation in nearly half of its members. A gene mutation is a permanent change in the DNA sequence that makes up a gene. Gene mutations occur by either being inherited or acquired (environmental). Mutations have the potential to have positive or adverse effects on the expression of a condition or disorder.

In addition to its role in the embryonic development of lung, heart, and intestinal tissues, the gene FOXP2 has been found to play a pivotal role in the embryologic development of neuromolecular pathways of the brain. Vernes, Oliver, Spiteri, Lockstone, Puliyadi, et al. (2011) recently found evidence that FOXP2 modulates plasticity and connections of neuronal network formation by regulating mRNA (messenger RNA is a key molecule in translating DNA's genetic code into amino acids that make up proteins). There is also strong evidence that FOXP2 is involved in motor control of neural structures involved in sequential learning, planning, and movement of structures of the vocal tract (Fisher, Lai, & Monaco, 2003; Fisher & Scharff, 2009; Lai, Fisher, Hurst, Levy, Hodgson, et al., 2000). FOXP2 has also been implicated in the sequential constructs of neurolinguistic skills in language functioning (Leigois, Baleweg, Connelly, Gadlan, & Mishkin, 2003; Lewis, Shriberg, Freebairn, Hansen, Stein, et al., 2006). Constructs such as syntax, morphology, and phonologic processing have been associated with functions of FOXP2 (Enard, Przeworski, Fisher, Lai, & Wiebe, 2002; Vernes, Newbury, Abrahams, Winchester, Nicod, & Groszer, 2008). The exact nature of FOXP2's role in speech and language functioning however, is not well defined, and continues to be an object of interest and research.

Genetic research in the KE family revealed that FOXP2 mutation was a single gene, autosomal dominant mutation (50% inheritance rate) that had been inherited over four generations (Zimmer, 2011). Thirteen members of the KE family presented with a mutated FOXP2 gene on chromosome 7q31 (OMIN 605317) (Fisher et al., 2003; Hurst, Baraitser, Auger, Graham, & Norell, 1990; Lai, Fisher, Hurst, Vargha-Khadem, &

Monaco, 2001; Legeois, Baldeweg, Connelly, Gadlam, Mishklin, &Vargha-Khadem, 2003; MacDermott, Bonira, Sykes, Coupe, & Lai, 2005; Vargha-Khadem, Watkins, Alcock, Fletcher, & Passingham, 1995).

The affected family members exhibited severe verbal and oral apraxia (unique speech disorders involving the control and sequencing of muscles for speech) and difficulty in both verbal and written language skills (Feuk, Kalervo, Lipsanene-Nyman, Skaeeg, Nakabayashi, et al., 2006; Newbury & Monaco, 2010; Watkins, Donkers, & Vargha-Khadem, 2002).

Similar speech-language phenotypes (behavioral expression of many genes) have been found to occur with slight changes to the FOXP2 genotype (molecular expression of a gene) (Hurst et al., 1990; Lai, Fisher, Hurst, & Vargha-Khadem, 2001; Lai, Fisher, Hurst, Vargha-Khadem, & Monaco, 2001; MacDermott et al., 2005). In all likelihood, the speech and language phenotypic characteristics found in the KE family represent an exceptional, rare syndromic form (Feuk et al., 2006; Newbury & Monaco, 2010) rather than a distinct phenotype of a FOXP2 gene mutation (Feuk et al., 2006; Newbury & Monaco, 2010; Vernes et al., 2008; Zimmer, 2011).

The discovery of FOXP2 has also led to ongoing discoveries that have changed our understanding of the role of environment and possible candidate genes (a gene located on a chromosome region suspected of being involved in the expression of a trait of a disorder or disease). Discovering the contribution of genes to a phenotype also permits a better understanding of the contribution(s) of environment(s) leading to changes in how we view behavior.

There are currently five candidate genes associated with speech and language disorders that have been identified as a result of the discovery of FOXP2; CNTNAP2-GNPTAB (Fedyna, Drayna, & Kang, 2010), ROBO1, DCDC2, KIAAO319 (Rice, Smith, & Gayan, 2009). These candidate genes have brought new understanding of the neural substrates, genetic, and environmental relationship to specific disorders such as stuttering

(Andrews, Morris-Yates, Howie, & Martin, 1991; Felsenfeld, Kirk, Zhu, Statham, Neale, et al., 2002; Kang, Riazuddin, Mundoff, Krasnewich, & Friedman, et al., 2010), voice (Gray & Thibeault, 2002; Gray, Thibeault, & Ylitalo, 2009; Thibeault, Gray, Li, Ford, Smith, & Davis, 2002), speech sound disorders (Fisher, Vargha-Khadem, Watkins, Monaco, & Pembrey, 1998; Iyengar, 2003; Shriberg, Ballard, Tomblin, Duffy, Odell, & Williams, 2006; Shriberg, Tomblin & McSweeny, 1999), apraxia (Lewis, Shriberg, Freebairn, Hansen, Stein, Taylor & Iynegar, 2006; Shriberg, 1993, Shriberg & Austin, 1998), pediatric and adult language development and disorders (Barry, Yasin, & Bishop, 2007; Bishop, 2000, 2001; Bishop, North, & Donlan, 1995; Brzustowicz, 1996; Falcaro, Pickles, Newbury, Addis, Banfield, et al., 2008; Pennington & Bishop, 2009; Rice, Smith, & Gayan, 2009; SLI Consortium, 2004; Tomblin, Shriberg, Williams, Murray, & Patil, 2009), swallowing disorders (Hu, Preston, Post, White, & Kikuchi, et al., 2000; Mennella, Pepinon, & Reed, 2005; Orenstein, Shalaby, Whitcom, & Baramada, 2002; Orenstein, Whitcomb, & Barmada, 2005; Post, Ze, & Ehrlich, 2005), and hearing disorders (Dror & Avraham, 2010; Hilgert, Alasti, Dieljens, Pawlik, Wellnik, et al., 2008; Resendes, Williamson, & Morton, 2001).

The discovery of FOXP2 has also launched increased attention and research in complex trait disorders that often interface with communication disorders. As a result, researchers are seeing new levels of understanding of the relationships between genotypic and phenotypic signatures and neurodevelopmental constructs such as cognition, learning, reading, literacy disorders (Scerri & Schulte-Karen, 2010; Stein, 2004), autism (Autism Genome Project Consortium, 2011; Geschwind, 2011; Sousa, Clark, Holt, Panamanian, Mulder, Minder, et al., 2010), and behavioral-temperament traits (such as stress, attention, resilience, motivation) (Abrahams & Geschwind, 2010; Fisher, Francks, McCracken, McGough, & Marlow, 2002; Newbury & Monaco, 2001; Richardson, 2004; Seong, Shimizu, Nakamura, & Ishii, 2011; Zhou, Dempfle, Arcos-Burgos, et al., 2008).

Impact on Communication Sciences

Although the full extent of the genetic and genomic relationships to specific disorders has yet to be fully revealed, there is sufficient information available to challenge our perceptions of genetics and have direct impact on patient care. It is becoming increasingly recognized that advances in the understanding of genetic pathogenesis are leading to changes in theoretical concepts and clinical applicability. As stated by the American Speech-Language Hearing Association (ASHA) and the National Coalition for Health Professional Education in Genetics (NCHPEG) – a coalition of more than 150 organizations – "the manner in which individuals with communication disorders are diagnosed, assessed, and managed is being reexamined" (NCHPEG, 2004, p. 2; see also ASHA, 2004, 2006; NCHPEG, 2007). Concepts in the etiology of communication disorders, their classification system, and knowledge of a disorder's natural history (prognosis) are being challenged. Changes in theoretical concepts will impact the assessment, management, and treatment of communication disorders.

Etiology

The etiology of communication disorders has traditionally been associated with environmental and biological factors. The biological approach includes structural and neurological effects. For several decades communication disorders secondary to biological factors have been known to be associated with syndromes (single-gene disorders), modest to high familial concordance and heritability rates (Bishop et al., 1995; Lewis, 1992; Shriberg & Austin, 1998; Simberg, Santtila, Vajonem, Sala, & Sandnabba, 2009; Tomblin & Buckwalter, 1998; Tomblin, Records, Buckwalter, Zhang, Smith, & O'Brien, 1997). A genetic relationship was often suspected, however had not begun to be investigated until the start of the genomic age. Since then, biological and environmental models of the pathogenesis of communication disorders have been re-examined.

Shriberg (2010) suggests the "*polygenic-environmental causal model"* (PECM) as further delineation of biological approaches to understanding pathogenesis in communication disorders. This model is based on newly established quantitative phenotype-genotype associations resulting from genetic research conducted in the last ten years. A polygenic-environmental causal model includes interaction effects of genetics, genomics, gene x environment, and epigenetics (the effect of the interaction of gene x gene). In other words, multiple common variants (genetic and environmental), of varying effect sizes (small to moderate), interact with each other across time to manifest as individual genotypic and phenotypic traits. These traits contribute to normal variation in human behavior. Yet, these trait variants also increase the susceptibility of a disorder or a condition for many others.

The role of genetics in speech and language disorders is now widely accepted in the scientific community, as witnessed by the inclusion of such disorders in the Mendelian Inheritance in Man database. The OMIM includes entries for several communication disorders, acknowledging the genetic etiology for speech sound disorders, dyspraxia (also known as apraxia), stuttering, speech language disorders, and specific language impairments (see Table 1).

Improved understanding of genetic pathogenesis has suggested redefinitions for types and subtypes of many communication disorders and their co-morbid conditions (Tyler, 2010; Lewis, 2010). For example, it is well known that first-degree relatives of the subject being affected or studied with autism spectrum disorder (ASD) present an increase in social and language dysfunction, usually presenting with less severity of expression. This expression of phenotype is called *the broader phenotype* (Ben-Yizhak, Yirmiya, Seidman, Lord, & Sigman, 2011). Studies of multiple measures of sub-threshold traits suggest features of a quantitative continuum of function that is inherited in distinct patterns (Ronald, Larsson, Anckarsater, & Lichtenstein, 2010; Steer, Golding, & Bolton, 2010).

Attention is also being directed at new perspectives on the etiology of stuttering to further delineate etiological subtypes based on phenotypic and genetic expression (Yari & Ambrose, 2002). Several recent several studies have provided evidence for at least three genes associated with chromosome 12: GNPTAB, GNPTG, NAGPA (OMIM) (Kang et al., 2010; Suresh, Ambrose, Roe, Pluzhnikov, Wittke-Thompson, & Wu, et al., 2006; Yari & Ambrose, 2002). Researchers have suggested various etiologic subtypes specific to phenotype expression and certain cytogenic (chromosomal) locations.

Table 1. ***OMIM Database for Speech, Language and Stuttering Disorders***

Phenotype OMIM #	Disorder Type	Gene/ Locus	Cytogenetic Location
%184450	Speech Sound Disorder	*	3q12-q13
#602081	Speech Language Disorder	SPCH1	7q31.1
%606711	Specific Language Impairment 1	SLI1	16q
%606712	Specific Language Impairment 2	SLI2	19q
%607134	Specific Language Impairment 3	SLI3	13q21
%612514	Specific Language Impairment 4	SLI4	7q35-q36
#613670	Mental Retardation with Language Impairment and Autistic Features	*	3p13
%184450	Stuttering, Familial Persistent 1	STUT1	18p11.3-p11.2
%609261	Stuttering, Familial Persistent 2	STUT2	12q24.1
#3006433	Rolandic Epilepsy, Mental Retardation, and Speech Apraxia-X Linked	RESDX	Xq22.1
%601085	Rolandic Epilepsy, Mental Retardation, and Speech Apraxia – Autosomal Dominant	RESDAD	7q31.1

*Undetermined # Known Phenotype % Unknown Phenotype

Phenotypic variant factors such as age of onset, metabolic function, co-morbid presentation with language impairment, and/or speech sound disorders, response to

recovery, and response to therapeutic remediation have been implicated as indices of possible identifying etiological substrates of stuttering disorders (Kang et al., 2010; Suresh et al., 2006; Yairi & Ambrose, 2002).

A similar trend is also occurring with respect to speech sound disorders. Various phenotypic expressions of speech sound disorders (isolated articulation disorder versus speech sound disorder, or speech sound disorder accompanied by language disorder and/or reading disorder, etc.) are challenging our perceptions of causation, inclusionary and exclusionary diagnostic criteria, and varied responses to therapeutic remediation (Shriberg, Lewis, Tomblin, McSweeny, Karlsson, & Scheer, 2005).

Although there is a strong relationship between genetics, environment, and epigenetics (heritable alterations in gene expression caused by mechanisms other than changes in DNA sequence) in many communication disorders, we continue to face the challenges inherent in definitive delineation of etiology. Genetic heterogeneity, complex genetic models with many contributing loci of varying effects, gene-by-gene interactions, and gene by environment interactions are some of the issues adding to the difficult process of etiological delineation for complex trait disorders. Yet, knowledge of possible genetic causes of a disorder or disease allows appropriate diagnostic tools to be used in the assessment of an individual.

Classification System

Diagnostic categories that were largely based on behavioral observations are now being validated by genetic information, resulting in new classifications of phenotype-genotype constructs (Lewis, 2010). These data suggest that different features of a disorder (e.g., language disorders) represent a quantitative continuum of phenotypic and genotypic manifestations resulting from the interactions of genes, genomics, and environment (Geschwind, 2011). This is supported by the fact that specific genetic factors contribute to the development and function of specific brain structures, and the

hypothesis that distinct brain circuits might underline different constructs of a communication disorder (Enard et al., 2005**;** Newbury & Monaco, 2010). Genetic research has provided evidence for subtypes (diagnostic markers) of speech sound disorders, language impairments, and reading disorders (Smith, 2007; Smith, Pennington, Boada, & Shriberg, 2005; Stein, Millard, Kluge, Miscrimarra, Cartier, Freebairn, et al., 2006; Stein, Schick, Gerry-Taylor, Shriberg, Millard, et el., 2004), This research has also provided insight into the etiological mechanisms implicated in speech sound disorder and specific language impairment, cognitive overlap of co-morbid disorders such as reading, spelling, autism, attention deficit hyperactivity disorder (ADHD), and learning disabilities. Whether one views classification of disorders as bimodal (affected or unaffected) versus a quantitative spectrum of impairments, often with co-morbid presentations (Geschwind, 2011), it is important to discover the etiology and the presentation of the behavior to fully understand how we may assess, manage and research.

Narrower classifications of diagnostic markers of phenotypes based on a polygenetic-environmental model have redefined categories into both phenotypic and genotypic subtypes. Redefining the classification system allows for specificity in the identification and verification of susceptibility risk rates of occurrence and recurrence.

Prognosis

Genetic advances have broadened our understanding the natural history of a disorder. Natural history is the description of the uninterrupted progression (not treated or manipulated) of a disorder or disease. Knowledge of natural history is as important as knowledge of the etiology. For example, epidemiological studies have found that in certain types of speech sound disorders, children will also present with reading difficulties; whereas in other subtypes, they may present with sound disorders, language impairment and reading difficulties.

Studies have shown that remediation and therapeutic progress is dependent on the subtype and the age at which therapeutic intervention began as well as duration of intervention. For example, we are beginning to understand more about how stuttering may manifest over time. Pediatric onset stuttering may spontaneously resolve by five or six years of age, whereas stuttering with an acute onset in childhood may not resolve (with or without therapeutic intervention) and may continue well into adulthood (Yari, Ambrose, & Cox, 1996).

There are postulations that these two groups are distinct from each other, exhibiting different genotypes and phenotypes. Information provided over the life span of an individual provides the "natural history" of one or several types of speech sound disorders. Having knowledge of the natural history therefore provides information on how a disorder may manifest over time as the subject ages, with different treatment modalities, and differing interactions between genes and the environmental contexts. Based on familial, environmental and genetic information, specific prognostic statements with regard to susceptibility to a speech language disorder, as well as the risk for occurrence and/or re-occurrence, can be made with increased certainty.

Assessment, Management, and Treatment

The current wealth of genetic and genomic information has clinical applications for the assessment, diagnosis and management of communication disorders. Advances in our understanding of the genetic pathogenesis of a disorder allow for a broader definition of possible etiologies. In turn, understanding of the involvement of genetics in communication disorders has called into question our classification system and definitions of communication disorders.

New understanding is leading to changes in assessment paradigms, tools and techniques. Obtaining information on the patient's family history in addition to pedigree analysis allows identification of specific biologic- genetic markers or "bio-gen markers,"

providing information on genetic susceptibility, occurrence and recurrence (see Table 2). Physical assessment including identification of possible patterns of structural deviations from normal contributes to identification of bio-gen markers. Identifying the risk of genetic communication disorders based on presenting phenotypes with the assistance of a screening tool (composite of bio-gen markers) will improve on diagnosis and management.

Table 2. ***Bio-Gen Markers for Genetic Susceptibility of Genomics***

Tools	Bio-Gen Markers
Family information	Malformations of one or multiple organ systems
	Multiple family members affected
	Presence of conditions in the less often affected gender (e.g., breast cancer in males or persistent stuttering in females)
	Presence of overlapping symptoms (specific language impairment (SLI), dyslexia, and stuttering)
	Abnormalities in growth
	Recurrent pregnancy losses
	Close biological relationship to parents
	Ethnic predispositions to certain genetic disorders, diseases, conditions. History of therapeutic interventions for neuro-developmental disorders
Physical examination	Identify structural anomalies that may contribute and or present as co-morbid factors to the identified communication disorder, and/or present with patterns that may identify a syndrome relationship.
Predictive screening tools	Administer screening tools, which provide a composite score of type and severity of genetic communication disorders and or co-morbid conditions, family history and pedigree risk, and physical patterns.

Genetic subtypes of disorders may present with distinctive phenotypes dictating a particular course of therapy (Shriberg, Flipsen, Karlsson, & McSweeney 2001; Shriberg, Lewis, Tomblin, McSweeny, Karisson, & Sheer, 2005). Pharmacological management of stuttering is not new; previously, selective serotonin re-uptake inhibitors (SSRI) were used to treat the manifestations of stuttering. SSRI's are a class of compounds typically

used in the treatment of anxiety and depression disorders. With recent genetic findings, pharmacological treatment of lysosomal storage deficiencies in some stuttering individuals with a specific gene mutation may be a reality in the near future. Suspecting and/or knowing a genetic cause of a speech and language disorder may provide further medical and legal evidence for an individual to receive appropriate management in cases where the issues are not well understood or yet manifested.

For disorders such as speech-language impairment, it is hoped that the information from genetic research in combination with speech-language pathology, psycholinguistic and neurological data, will aid in the development of better predictive test batteries, thus allowing for early identification and better treatment of those individuals at risk.

How genetics will directly impact treatments of various communication disorders has yet to be fully realized. Many complex diseases involve interactions among multiple genes and multiple aspects of the environment. It seems reasonable, however, to define genetic risk through thorough, well-defined bio-gen markers. Clearly, relevant bio-gen markers are needed. They are predictors with the highest possible fidelity based on our current understanding of the evidence that could foretell the likely long-term clinical effects of behavioral interventions.

Identification of bio-gen markers facilitates enhanced management of patients in regards to appropriate referrals to geneticists for testing and at times genetic management (medical, pharmacological, and gene therapy). Advances in technology and genetic screening will facilitate early identification of speech and languages issues. This leads to improved monitoring and allows for the initiation of early intervention. Therapy techniques used in the field of speech- language pathology may be tailored to fit individual differences associated with genetic subtypes and the resultant underlying deficits.

Genetics and Genomic Recognition in Communication Sciences

The recognition of the significance of genetic information in communication sciences is noted in the increased number of articles with genetic and genomic focus published by the *Advance- Speech and Audiology.* The *Advance,* with a circulation of over 61,000 readers is a biweekly publication for speech language pathologists (Merion Publications, 2008). In the years between 1994 and 2011, a total of 462 articles with specific genetic subject matter were published in periodical (Table 3). The number of genetic related articles published during this time indicated an increasing trend. A significant increase in publications was noted with the release of the working draft of the Human Genome Project in 2000 (18 articles) and the publication of the completed HGP in 2003 (39 articles) with a peak in 2008 with 59 articles published with genetic and genomic content.

Table 3. ***Number of ADVANCE Publications with Genetic and Genomic Content***

Year	No. of Articles	Year	No. of Articles
1994	5	2003	39
1995	4	2004	39
1996	3	2005	25
1997	6	2006	39
1998	8	2007	48
1999	12	2008	59
2000	18	2009	40
2001	15	2010	39
2002	18	2011	45
		TOTAL	462

Research in communication sciences have been influenced by the advances in genetics and genomics. ASHA conducted the *2008 Researcher Survey* in an effort to better serve the interests and needs of the communication sciences and disorders research

community. The survey asked researchers in communication sciences questions regarding the general type and area of research they had engaged in from the years 2003 to 2008. A total of 1,233 surveys were sent with 303 responses received (response rate of 24.6%).

The respondents were asked to indicate the focus of the research that they had conducted over the past five years (multiple responses were allowed). More than half reported that they were involved in clinical research specific to the nature of a disorder (56%), assessment/diagnosis (53%), and/or treatment (53%). A slightly lower percentage (45%) indicated that basic research had been their primary area of focus, and only 11% reported that their research focused on clinical research in the area of prevention (ASHA, 2008).

The respondents were also presented with a list of 64 possible areas of research interest. Respondents indicated that at least 7.9% of research was in the area of genetics (see Table 4). Genetics research received relatively the same percentage of research interest as research in schools (7.9%), closely followed by research in swallowing disorders (7.6%) and voice disorders (7.6%). The areas receiving the highest percentage of responses were language disorders (36%), language acquisition (28%) and language (24%), while the lowest percentages were noted in head neck cancers/laryngectomy (1.3%), intraoperative monitoring (1.0%), and myofunctional disorders (0.3%; see Table 4).

Recognizing that there is a role for knowledge of genetics and genomics in speech-language pathology, the American Speech-Language Hearing Association (ASHA) in conjunction with the National Coalition for Health Professional Education in Genetics (NCHPEG) – a coalition of more than 150 organizations – developed a Web-based resource called *Genetics in the Practice of Speech-Language Pathology and Audiology*. The website provides genetics instruction and resources for speech-language pathologists.

Table 4. ***Percentage of Genetic and Genomic Research in Comparison to Area of Interest***

Area of Research Interest	Response Percentage	Area of Research Interest	Response Percentage
Language disorders	36.0%	Second language acquisition	8.9%
Language acquisition	27.7%	Auditory/vestibular physiology	8.3%
Language	23.8%	**Genetics**	**7.9%**
Normal processes	22.4%	Service delivery in the schools	7.9%
Literacy	21.8%	Swallowing disorders	7.6%
Hearing	19.8%	Voice	7.6%
Neurogenic communication disorders	19.1%	Augmentative/alternative	7.3%
Aging	18.5%	Hearing conservation	6.9%
Learning Disabilities	17.5%	Childhood apraxia of speech	6.3%
Cochlear Implants	16.2%	Anatomy	5.9%
Cultural and linguistic variables	15.2%	Developmental speech motor control	5.9%
Diagnostic/clinical processes	15.2%	Fluency	5.6%
Psycholinguistics	14.2%	Dementia	5.3%
Cognitive-communication	14.2%	Otoacoustic emissions	5.0%
Speech disorders	14.5%	Acquired apraxia of speech	4.6%
Hearing disorders	13.9%	Universal newborn hearing screening	4.3%
Aphasia	13.5%	Oral motor development	4.0%
Psychoacoustics	13.5%	Tinnitus	3.3%
Articulation/phonological disorders	13.2%	Cleft palate	3.3%
Speech motor control	13.2%	Balance disorders	3.3%
Dyslexia/reading disabilities	12.9%	Supervision	3.0%
Neurolinguistics	12.5%	Brain stem implants	2.6%
Speech	12.5%	Electrocochleography	2.6%
Traumatic brain injury	11.9%	Assistive listening devices	2.0%
Electrophysiology	11.9%	Oral motor training	2.0%
Audiologic (re)habilitation	10.2%	Accent reduction	1.7%
Physiology	10.2%	Feeding disorders	1.7%
Aural (re)habilitation	9.6%	Hair cell regeneration	1.3%
Central auditory processing	9.6%	Head and neck cancers/laryngectomy	1.3%
Speech acquisition	9.6%	Intraoperative monitoring	1.0%
Hearing disorders	8.6%	Orofacial myofunctional disorders	0.3%

(Adapted from 2008 ASHA Researcher Survey Section Report: Areas of Research)

The voluntary, web-based program (http://www.nchpeg.org/shla/site.html) is based on current genetic principles, and primarily describes Mendelian genetics as it relates to speech-language pathology.

In 2007, NCHPEG in collaboration with ASHA developed core competencies for speech- language pathologists called the *Core Competencies in Genetics Essential for All Speech-Language Pathologists* (NCHPEG, 2007). The goal was to ensure that all health professionals including speech-language pathologists have basic standards by which to begin designing and creating curricular content for genetic education (Kanny, Smith, & Dundgen, 2005). Although these competencies have been developed, they are not specific to the field of communication sciences, have not been validated, and have not been well disseminated to the membership of professional speech-language pathologists and training programs (LeBlanc, 2010).

The American Speech-Language Hearing Association has published articles on genetic related topics in the *ASHA Leader,* with regards to genetics in clinical practice (ASHA, 2010; Coufal & Schaefer, 2003). ASHA has offered genetic information on its website (ASHA, 2011) and has offered genetically-based content in its annual meetings (Garret, Harvey, Neils-Stunjas, Steinberg-Warren, & Lewis, 2006; Lewis, 2008).

Genetic Knowledge in the Field of Communication Sciences

With new information from genetic research, come new demands on the speech-language pathologist. They are often the first professional to see individuals with genetic speech or hearing conditions (Lapham et al., 2000). “Speech-language pathologists regardless of specialty area, role, or practice setting, will face questions about the implications of genetics from their patients” (NCHPEG, 2007, p. 2). Not only does this new information have a direct relationship on the scope and breadth of practice

management of individuals with communication disorders, it reflects issues of professional preparedness of the field of speech language pathology.

As early as 1993, speech-language pathologists indicated they lacked general knowledge about genetics, genetically-based conditions and genetic counseling. Chermak & Wagner-Blitz (1993) in a survey to determine the perception of genetic knowledge of 300 speech-language pathologists found that 79% of the respondents perceived their knowledge of genetics to be “marginal to none.” The survey also asked questions related to the knowledge of genetic principles (what was known of genetics in the pre-genomic era). The performance of speech- language pathologists, at 23% mean correct responses, was statistically lower ($p<.05$) than that of audiologists (53% mean correct responses). Lapham et al. (2000) conducted a random sample survey in 1998 of members of six professional associations (American Dietetic Association, American Psychological Association, American Physical Therapy Association, the American Association of Social Workers, and the American Speech- Language and Hearing Association). Results indicated that all respondents from each association described their perceived genetic knowledge as “low”. In 2000, Willig, Moss, and Lapham reported that more than 80 percent of professional speech-language pathologists surveyed continued to report little to no background or education in genetics.

In a follow-up publication of the same study, Long, Brady, and Lapham (2001) reported on the demographics, work settings, client characteristics, and provision of genetic services, job responsibilities, and genetic education of speech-language pathologists. Seventy-one percent of speech pathologists surveyed indicated they discussed a genetic component of a presenting clinical problem with the patient, while a genetic referral was made by 26% of the respondents. Twenty-five percent indicated they provided counseling about genetic concerns, while 10% indicated they provided counseling to clients making decisions about whether to have genetic testing. Yet, only 11% reported being confident in discussing the genetic basis of disorders or conditions.

In over ten years since the initial survey on perceptions of genetic knowledge of professional speech language pathologists, Christianson et al. (2005) reported that between the years 1997 and 2002, 79%of speech-language pathologists continued to describe their knowledge of basic genetic science as “marginal to none”. In spite of the fact that they acknowledged “poor knowledge” of genetics, 43% of respondents reported discussing with families and patients at least one of eight genetically related topics. Lack of knowledge was demonstrated in areas relating to means of genetic transmission; techniques used for prenatal diagnosis; genetic conditions involving speech, language, or hearing disorders; and the indications for and objectives of genetic evaluation and counseling.

These perceptions have existed prior to the beginning of the “genetic informational age” and persist well into the post-genomic era, in spite of the public awareness efforts of the mass media, National Institutes of Health (NIH), NCHPEG, and ASHA. Although there is the recognition of a role for genetics and genomics in the field of speech-language pathology, there remains inadequate translation to the professional speech language pathologist. The fast pace of genetic advances, and “the paucity of professional training, leaves many providers without up-to-date answers for their patients” (NCHPEG, 2007, p. 2). We are facing challenges on how to translate genetic advancements in the genetic education of speech-language pathologists.

Reasons for Poor Genetic Knowledge/Literacy

There are several reasons why the field of speech-language pathology may not be meeting the demand for better education in genetics. Genetic and genomic information outside the genetic community is not well understood and has not been well disseminated (NCHPEG, 2007). As Geller, Bernardt, and Holtzman (2002) claim, “the mass media are primary sources of health and science information for many Americans, including scientists and physicians” (p. 773; see also Conrad & Markens, 2001; Young, 2002). As

Long et al. reported in 2001, most members of allied health professionals obtained information about genetics from the media: newspaper and magazines (83%); television and radio, (76%); professional journals or books (74%), and conferences (32%). Twenty-five percent of respondents reported to have received information from their patients.

The relationship between genetics and communication sciences has not been well translated to clinical practice. Genetics was traditionally viewed simplistically as primarily a Mendelian relationship, meaning inherited transmission. The role of genetics was typically considered to be associated with specific populations, namely clefting disorders and syndromes. Although familial inheritance was considered as a possible contribution to certain types of communication disorders, it was not thought to have a vital impact on decisions regarding assessment and or management in speech-language pathology (Shprintzen, 2001). Consequently knowing a "genetic diagnosis" minimally affected how the speech-language pathologist assessed, diagnosed, managed or treated communication disorders.

Although there are increasing initiatives to inform the profession of speech language pathologists, the ASHA continuing education policies are specifically directed at the professional speech-language pathologist, not towards the speech-language pathologist in training (undergraduate and graduate degree programs). Continuing education is a requirement to maintain certification with ASHA and maintain respective state licensure (ASHA, 2011). Students do not receive continuing education credits or recognition for attending continuing education events. Therefore, students do not have access to the same genetic education opportunities as professional speech-language pathologists. The only opportunities where a student may receive genetic and genomic information regarding communication sciences are in undergraduate and or graduate degree programs. However, it is unknown whether genetic and genomic related content is offered in the speech-language pathology undergraduate or graduate curricula.

Genetics may not have had a prominent place in speech-language pathology curricula because it is suspected that faculty teaching in speech-language pathology programs received their basic science education in the pre-genomic era. It is highly likely that current faculty members are lacking in genetic literacy, especially genetic and genomic principles directly related to communication sciences. It is suspected that speech-language pathologists are not being trained in undergraduate and graduate programs in areas of basic genetic science and its direct applicability to communication sciences. To date, no known literature has been published on genetic related content in the curricula of academic speech-language pathology degree programs.

Curriculum requirements for speech-language pathology degree programs have been established by the Council on Academic Programs in Communication Sciences and Disorders (CAPSCD) and the Council on Academic Accreditation in Audiology and Speech-Language Pathology (CAA). Their role in part, is to formulate standards for the accreditation of graduate education programs and to provide accreditation to audiology and speech-language pathology programs. The CAPSCD standards are based on and approved by the Council for Higher Education Accreditation (CHEA) and U.S. Department of Education (USDE). A review of the curriculum standards indicates broad statements providing the accommodation for inclusion of “biological sciences,” yet it does not provide specific inclusion regarding genetics and genomics (The IIIB Standards for Accreditation of Graduate Education in Audiology and Speech-Language Pathology (ASHA, 2012).

> 3.3B The scientific and research foundations of the profession are evident in the curriculum.
> The program must demonstrate how it verifies that students obtain knowledge in the basic sciences (e.g., biological, behavioral, physical science, and mathematics), basic science skills (e.g., scientific methods and critical thinking), and the basic communication sciences (e.g., acoustics; physiological and neurological processes of speech, language, and hearing; linguistics). The curriculum must provide opportunities for students to become knowledgeable consumers of research literature with an emphasis on

> the fundamentals of evidenced-based practice, as well as the application of these principles and practices to clinical populations. The curriculum must reflect the scientific bases of the professions and include research methodology, research literature, and opportunities to participate in research and scholarship activities, consistent with the mission and goals of the program, institution, and profession.

The standards include a list of content topics, which should be included (such as voice, stuttering, and articulation). Noticeably absent from the standards are genetically and genomic sciences. In turn, the respective speech-language pathology university program is afforded the autonomy to develop curricula as long as it includes the standard requirements as per CAPSCD, ASHA, the CHEA, the USDE, and abide by respective State standards.

Although there are currently no requirements for undergraduate and graduate degree programs to include genetic and genomic content (basic science principles and discipline specific subject matter) into their speech-language pathology curricula (CAPCSD and the CAA, 2011), approximately 20 percent of the Praxis Series included genetic-related topics (Garrett, Neils-Strunjas, Steinberg, Warren & Kishman, 2005). Thirty-one percent of the questions in audiology relate to genetics in the Praxis Specialty Test (ASHA, 2004). The Praxis Specialty Test is a mandatory examination of ASHA certification required of all speech-language pathologists.

A possible reason for the exclusion of genetic information in curricula is that it is only in recent history that genetics and genomics have a redefined presence and have established roles in the field of communication sciences. In review of ASHA's policies, position statements, and ad hoc committee statements on roles and responsibilities of the speech-language pathologist, there are no known formal positions offered by ASHA on genetics and/or genomics with regards to inclusion. A similar finding was found when searching position statements on requirements for graduate training programs. To the best of knowledge there are no known inclusion statements specific to genetics and or genomics. This speaks to the fact that governing agencies setting curriculum policy, and

degree program chairpersons, directors and faculty members may not be fully aware of the advances made in genetics and genomics and its impact on the field of communication sciences. A literature search indicates there are no known publications focusing on what is known about genetics and genomics authored by those making policy standards and by those developing standard curriculum requirements.

In addition, operational definitions of what it is to be "genetically or genomically literate" have not been established for the specific needs of the speech-language pathology profession (LeBlanc, 2010). There are various definitions of genetic literacy in the literature (Andrews, Fullarton, Holtzman, & Motulsky, 1994; McInerney, 2002; Vice Bowling et al., 2008), but these generally apply to medical professionals and are not "discipline specific." A review of the ASHA position statements on the roles and responsibilities of speech-language pathologists with regard to genetics and genomics have not been developed to date.

Promoting the Integration of Genetics

The field of speech-language pathology is in need of re-conceptualizing the role of genetics and genomics in order to be able to further translate the sciences of genetics and genomics to the pathogenesis of communicative diseases and disorders. Acknowledgement of the presence and role of communication and its disorders in genetics is not enough and does not address how this will translate into clinical practice. Speech-language pathologists need to *"think genomically"* and possess competencies in knowledge and skills for safe and effective clinical practice. Yet, in spite of the genetic information currently available and its direct relevance to the field of speech-language pathology, very little is known about the attitudes, beliefs and experiences of practicing speech pathologists and educators. Meeting the educational needs of professionals and facilitating new trends in curricula of undergraduate and graduate students in the field, remains a challenge.

In review of the literature, several key issues were revealed: (1) recent genetic and genomic advancements have a role in communication sciences, (2) speech-language pathologists are interested in receiving more information about genetically related communication sciences, yet feel they are poorly trained, and (3) there are many unanswered questions about genetics and genomics in the field of speech-language pathology and communication sciences. The questions listed in Table 5 encompass three major areas-- professional issues, higher education, and national policy standards and regulations in regards to professional competency.

Table 5. ***Questions in Areas of Professional Issues, Higher Education and National Policy***

Professional issues	What do speech-language pathologists know about genetics, genomics and its impact on the field of speech language pathology? What are speech pathologists' attitudes toward genetics, genomics and its impact on the field of speech-language pathology? How are genetics and genomics being used clinically? How should post-genomic genetics be used in the clinical practice of speech-language pathology today? What criteria are currently being used by the speech-language pathologist for decisions regarding genetics?
Higher education	How is the speech-language pathologist becoming educated in genetics? What is being taught in the undergraduate and graduate programs in regards to genetics and genomics? How do we educate the field of speech-language pathology in regards to genetics and genomics? How is the field of speech-language pathology going to take a larger role in genetic research in the future?
National policy standards and regulations	What is genetic literacy in communication sciences? What are the discipline-specific professional competencies required in clinical practice? What are the roles and responsibilities (practice standards) for entry and maintenance as ASHA certified speech-language pathologists? What are the standard curriculum requirements genetic content for undergraduate and graduate degree programs?

Having to ask such questions with no immediate answers at hand reflects the current status of the field of genetic communication sciences. The field of speech-language pathology is challenged with translating and incorporating genetics and genomics into theoretical foundations of communication sciences, integrating the knowledge into clinical practice, and being coordinating standards with training programs in both the education of the practicing speech-language pathologist as well as the student speech language pathologist. Success with meeting these challenges begins with answering many of the questions raised. Integral to meeting the challenges of a translational process is a needs analysis of the current state of the field of communication sciences in relationship to genetics and genomic. This will not only provide answers to many of the questions raised, but will also provide a platform from which the science of genetics and genomics can further develop the field of communication sciences.

Evidence from a needs analysis will reframe the context of communication sciences more broadly to explore genetic and genomic properties of an individual's communication and disorder within a larger, integrated system.

A systematic review of professional issues and higher education will demonstrate that the clinical practices of the professional speech-language pathologist and the basic structure of the undergraduate and graduate curriculum in degree programs in speech-language pathology are not matched to the nature and pace of scientific and societal changes with regards to genetic and genomic advances. Such information will provide a foundation for professional dialog leading to changes in competency and curriculum standards for the speech-language pathologist at the national level.

Conclusion

In the past decade, remarkable strides have been made in genetics that have profound implications for healthcare education, research and clinical practice. Speech-

language pathologists as integral providers in the contemporary health arena must have a larger presence in the area of genetics and must become knowledgeable in relevant aspects of genetic medicine. By providing a platform of genetic knowledge and clinical skill to its members, the field of speech pathology can move towards the goal of becoming genetically literate to best serve the profession, its members, the research community, and individuals and families seeking clinical services.

Chapter III

METHODOLOGY

This chapter describes the methodology used for this investigative research. The theoretical framework of this study followed a process synthesized from approaches used in needs assessment and analysis research (Garvin-Doxas & Klymkowsky, 2007; Treagust 1988, 1995; Vice Bowling et al., 2008; Witkin & Altschuld, 1995). Witkin and Altschuld (1995) define needs assessment as "a systematic set of procedures undertaken for the purpose of setting priorities and making decisions about program or organizational improvements and allocation of resources" (p. 4). The priorities are based on need, which has been defined as a discrepancy between a target state and an actual state (Witkin & Altschuld, 1995). Witkin and Altschuld proposed a *Three-Phase Model Theory* of needs assessment. The theory is based on a framework of analysis, assessment and action procedures. These processes occur over three phases: pre-assessment (exploration), assessment (data gathering), and post-assessment (utilization), and provides a model of evidence for change. The result is the development of two survey instruments, which will address the aims of this research study. This study was granted exempt status by the Teachers College, Columbia University Institutional Review Board (IRB Approval #12-153) (see Appendix A).

The methodology and study design will be presented according to each of the two survey instruments used in this research, targeting (a) program directors' views of genetic

and genomic content in degree training programs, and (b) genomic education of professional speech-language pathologists.

Methodology and Research Design of Survey I: Program Directors of Degree Training Programs

Subjects

A survey was mailed to 242 program directors of ASHA-accredited undergraduate and graduate degree programs in speech-language pathology within the United States. The email addresses of degree programs were obtained from the Council of Academic Programs in Communication Sciences and Disorders (CAPCSD, September 2011) and respective academic program websites.

Methods

The survey instrument, *A Curriculum Survey of Genetic Content of Speech Language Pathology Degree Programs* and an accompanying cover letter (see Appendix B), were delivered to all academic programs by SurveyMonkey.com©, an online survey tool.

Program directors were informed that their participation would be voluntary with no incentive other than contributing to general knowledge. To facilitate a high response rate, program directors were initially notified of the survey by a pre-survey introductory letter (see Appendix C). The survey was subsequently sent to each prospective respondent three days upon receiving the introductory letter notification. One week after the initial mailing of the survey, non-responding program directors were contacted and surveys re-sent. This was repeated twice. The surveys were collected from February 10, 2012 to March 5, 2012. Data entry for returned surveys and statistical analysis were conducted using IBM SPSS v.19.0 (SPSS, Inc. Chicago, IL) software package.

Survey Development

The approach to designing the survey instrument used with the degree programs was similar to that used by others who have developed assessment tools in the behavioral sciences (e.g., Anderson, Fisher, & Norman, 2002; Hestenes, Wells, & Swackhamer, 1992; Hufnagal, 2002; Mulford & Robinson, 2002; Treagust, 1988, 1995; Vice Bowling et al., 2008).

The instrument was developed in several stages. Step 1 involved defining and development of key concepts to be integrated into content. Step 2 identified thematic concepts, query domains, and selection of survey items. Step 3 involved refinement of the survey by review by professionals, experts, and focus group interviews. Step 4 involved several strategies for validation of the final survey version. The specific steps utilized in survey development are outlined in Table 6.

Table 6. ***Steps Used in Survey Development***

Step	Methods Used in Survey Development	Description of Procedures
1	Extraction and validation of key concepts and query domains	1. Extractions supporting concepts from Essential Core Competencies in Genetics for all Speech Language Pathologists (NCHPEG, 2004, 2007). 2. Validate relevance to speech language pathology with experts. 3. Rank order concepts. 4. Select cutoff to establish 3 query domains and concepts.
2	Exploration and development of thematic concepts	1. Establish thematic concepts. 2. Develop Rubric of Genomic Education. 3. Apply content analysis.
3.	Pretesting and survey refinement	1. Administer draft survey to using cognitive, think-aloud interviews to check clarity of items, readability, and student reasoning. 2. Refine inventory based on interview findings.
4.	Pilot testing and survey revision	1. Administer pilot inventory 2. Analyze student Item and survey psychometrics 3. Reduce survey to beta version, retaining most robust items to cover key concepts.

Defining the Content and Selection of Items

Although establishing the content domain began during the initial survey development phase, the process was iterative and spanned all steps of inventory development as the domains and query items were repeatedly focused, refined, and reorganized.

Content domains were developed based on the questions raised from the literature review, benchmarks of genetic and genomic content identified by *Essential Core Competencies in Genetics for all Speech Language Pathologists* (NCHPEG, 2004, 2007), discipline specific concepts identified in the literature, and similar instruments used in medicine and allied health professions (Amos, Della Rocca, Karchmer, Culpepper, & Cohn, 2004; Thurston, Wales, Bell, Torbeck, & Brockaw, 2007).

Three query domains (or concepts) were defined: (1) perceived attitudes of genetics and genomics in communication sciences and disorders by program directors; (2) presence, and delivery format, of genetic and genomic content within program curriculums; and (3) thematic concepts of genetic and genomic content.

Query 1. Program Directors' Perceptions of Genetic and Genomic Content. Program directors were asked whether interest in genetics and genomics is expressed by both faculty and students (Items 19 and 20, respectively). The program directors' attitudes and opinions about genetics and genomics were probed, especially with respect to expected competences in knowledge and skill of graduating speech language pathologists (Item 7), and the roles played by genetics and genomics in various developmental constructs and communication disorders (Items 26 and 27). Table 7 outlines the query domains and concepts mapped according to item number and source.

Query 2: Presence and Delivery Format of Genetic and Genomic Content. Items in Query 2 were developed to ascertain the presence of genetic and genomic content in curricula (Item 8); to identify possible reasons for genetic and genomic content

not being offered (Item 9); and to determine whether there were future plans for incorporating

Table 7. ***Query Domains and Concepts Mapped to Item and Source***

	Query Domains	Item	Concept Mapping	Source
1	Program director characteristics	28, 32, 31	Length in position, areas of training, type of academic standing	NA
2	Program characteristics	1, 2, 3, 4, 5, 6	Types of graduate degrees offered, number of students	N/A
3	Attitudes of genetics and genomics in communication sciences	7, 19, 20, 26, 27	Perceived impact and role of genetics and genomics, challenges and barriers in teaching genetics content.	Chermak &Wagner-Blitz, (1993); Willig, Moss, & Lapham, (2000); Christianson, McWalter & Warren, (2005)
4	Presence and format of genetic and genomic content	8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20	Presence, amount in genetic genomic science and applied content, delivery format (type, instructional methods).	Core Genetic Principles in Genetics Essential for All Speech Language Pathologists (NCHPEG, 2007) Genetics Literacy Assessment Instrument for non-biology undergraduates (Vice Bowling et al., 2008) Statement paper, What does the speech-language pathologist or audiologist need to know about genetics when conducting assessments? (ASHA, 2006) Focus Group Interviews
5	Thematic concepts	7, 16, 17	Criteria of quantity and quality of genetic content, effectiveness of representing cognitive and practice related behavioral constructs required for genetic literacy and competency.	Council on Academic Accreditation in Audiology and Speech Language Pathology (CAA). Dimensions of Genomic Education in Speech-Language Pathology (Appendix G)

genetic content in the near future (Item 10). Descriptions of the structure of genetic content were solicited (Items 11, 12, 13, 14, and 15), and other items probed areas of study with genetic and genomic content (Item 16), types of genetic and genomic topics offered (Item 17), the manner of curriculum integration (Item 11), the qualifications of the faculty member teaching the content (Item 18), and the type of delivery method used to teach the genetic and genomic content (online, in person, or other instructional methods; Item 19). Item 21 requested information about the programs' engagement in genetically based communication sciences research.

Query 3: Determining Thematic Concepts. Thematic analysis was based on a rubric, Genomic Education Rubric, developed for the current study and fully described in Appendix D. Items 7, 16 and 17 were mapped to four cognitive and pedagogic dimensions integral to genomic literacy and competency. Table 8 provides an example of the rubric. The cognitive and pedagogic dimensions included: the Genetic/Genomic dimension, that represented the approach used (genetics (Pre-genomic) versus genomics (Post-Genomic); the Content dimension, that embodied the content of the approach (basic genetic science or the translational content related to the discipline of speech-language pathology); the Knowledge dimension, representing constructs of literacy (theoretical proficiency) or competency (proficiency in clinical application); and the Pedagogic dimension, that was focused on whether "awareness" knowledge (existence of a principle or concepts) as compared to "process" knowledge (practical knowledge, concerning the use of the principle or concept or skill) were emphasized.

These constructs, related to cognition and pedagogy, were included in the rubric because literature regarding genomic education in medicine and nursing has established the importance of problem-based and collaborative learning approaches (Charlin, Mann, & Hansen, 1998; Chinn & Malhorta, 2002; Goodwin, Miller, & Cheetham, 1991). Contextual relevance is key for the development of critical thinking skills for students who are presented with abstract concepts. The information in a genetics and genomics

curriculum may be perceived as relatively challenging; therefore, providing a practical context assists in gaining new knowledge and skill.

Table 8. ***Sample of the Rubric of Genomic Education Used in Thematic Analysis***

Item	Item Concepts and Constructs Used in Rubric for Survey I	Genetic/Genomic Dimension		Content Dimension		Knowledge Dimension		Pedagogic Dimension	
		Genetic	Genomic	Basic Genetic Science	Discipline Specific	Literacy	Competency	Conceptual	Process
7	Competencies expected of graduating students								
16	Courses in which genetic and genomic content are integrated								
17	Genetic and genomic topics integrated into content								

Content validity was established for the rubric that was used to evaluate survey items relating to genomic education. A review panel was convened to confirm face validity four constructs of the rubric. Four judges were selected for their knowledge in the area of speech-language pathology (2), genetics (1), and craniofacial-genetics related speech-language pathology (1). The judges were asked to rate each rubric construct using a four-point rating scale. The judges' ratings were quantified using the content validity index (CVI) proposed by Waltz and Bausell (1983). The CVI scale is shown in Table 9. Content validity was measured by the adequacy with which the test items sampled the content areas of: (1) genetic and genomic science principles; (2) discipline-specific applicability (science and clinical application) to the field of communication sciences; (3) constructs of literacy and competency; and (4) pedagogic constructs of conceptual and process knowledge. An overall content validity index (CVI) for the genetic content

survey was calculated (CVI =.817), revealing moderately high content validity. Content validity for all constructs is presented in Appendix E.

Table 9. ***The Content Validity Index (CVI)***

Criteria for Measuring Content Validity			
1. Relevance	**2. Clarity**	**3. Simplicity**	**4. Ambiguity**
1-not relevant	1-not clear	1-not simple	1- doubtful
2-need some revision	2- need some revision	2- need some revision	2 -need some revision
3-relevant but need some minor revision	3-clear but need some minor revision	3-clear but need some minor revision	3-no doubt but need minor revision
4-very relevant	4-very clear	4-very simple	4-meaning is clear

Adapted from Waltz and Bausell (1983)

Survey Structure and Format

The initial questionnaire began with a pool of 47 items, distributed across the query domains. All questions were created *de novo*, and underwent several modifications. The question formats were varied, and consisted of rating scales (e.g., Likert scales), split-folding questions (i.e., a general question that was followed up with clarifying questions), and inverted funneling questions (i.e., a closed question that was followed by a more general open-ended question). The survey instrument included both closed-ended and open-ended questions. Answers were not forced, so that the survey could be advanced if a question was unanswered. Several questions were formatted with skip logic, subsequently varying the sample size of the responded items. Free text areas at the end of questions solicited comments or suggestions about additional important concepts in that category. All text responses are provided in Appendix H.

Bias Reduction

Attempts were made to minimize biases of response, non-response bias, response set, and response order bias in the survey. Response bias occurs when respondents attempt to answer the questions in the way they believe the questioner wants them to respond rather than according to their true beliefs. Non-response bias arises from the fact that there are usually differences between the ideal sample pool of respondents and the sample that actually responds to a survey. Response set bias occurs when respondents do not consider each question and just answer all the questions with the same response (e.g., if respondents answer “disagree” or “no” to all questions). Response order bias occurs when a respondent loses track of all options and picks one that comes easily to mind rather than the most accurate.

Bias was minimized using several methods: by reversing the wording in some of the survey items, providing clear, precise, and short questions, eliminating loaded or leading questions, and avoiding the use of double-barreled questions and double-negative questions.

Establishing Survey Reliability and Validity

Representational Sample. A randomization procedure was not used in Survey I; therefore, the non-random sampling was validated by comparing whether selected characteristics of the population of interest (242 program directors) were represented in the sample.

Reliability. Analysis of homogeneity provides an indication of the internal reliability of the instrument. A Cronbach’s alpha was performed on items representing conceptual constructs. Any item with a Cronbach’s alpha of less than .700 was deleted from the survey. Internal consistency was assessed of thematic constructs of the rubric of genomic education (Items, 7, 16, 17). Cronbach’s alpha = .871 for genomic education rubric constructs. Results indicated high internal consistency.

Content Validity. Content validity refers to the extent to which the items on an instrument assess the same content, or how well the intended content material is sampled by the measure (Goodwin & Leech, 2003; Standards for Educational and Psychological Testing 1999). Content validity involves a rigorous process, such as using a panel of experts to evaluate the content validity of a measure. To establish content validity, this survey was initially developed from a content analysis of literature followed by a review process (focus group). This is similar to procedures used by Clemson, Fitzgerald, and Heard (1999). It was followed by further measuring content validity as an index as described by Walter and Bausell, (1983). The Content Validity Index (CVI) was obtained for the instrument by administering a pilot study. A description of the procedures used in the focus group and pilot study follows.

Focus Group (Cognitive Interview)

Cognitive interviews, much like focus groups, involve "the administration of draft survey questions while collecting additional written and verbal information about the survey responses" (Beatty & Willis, 2007, p. 287). Survey designers use interviews to identify variance in meaning by exploring clarity of items, readability, and respondent reasoning (Drennan, 2003). Interview findings inform revision of survey items to enhance instrument reliability and validity (Knafl et al., 2007).

The interviews used in the survey development employed a "think aloud" process. The interview consisted of eight judges, four professional speech-language pathologists and four speech-language pathology program faculty members. The focus group participants were asked to verbally report their comments, suggestions, and statements as they completed a draft survey. Helpful comments or suggestions for improvement were welcomed. Responses and information provided by the participants were collected. Additional direct questions (called probes) to elicit further information were used. The reviewers were asked to answer the following questions with "yes" or "no": "Does the

question reflect the query domain concept (or aim) of the research?" and "Is this a quality question?" See Appendix F for responses provided by the focus group.

Based on the results of focus group interviews, 15 items were deleted from the original pool of 47 items due to redundancy of concepts, length of questions, number of subscales, ambiguity of question format, and inappropriate concepts (e.g., one early question that related to molecular processes). Items indicating ambiguity, overlapping of questions, poorly constructed questions, and redundancy were revised. On the basis of the reviewers' responses to the questions and their individual comments, the items once again underwent revision.

Pretesting Survey: Pilot Study

As a means for further survey development, the revised survey (revisions made from the findings of the focus group) was administered to faculty members of four speech-language pathology masters degree programs in the New York area (one each from New York City, Queens, Westchester and Long Island). A copy of the survey was provided for each judge, and the purpose and objectives of the study were explained. The judges were asked to complete the survey and subsequently rate each question on a four-point scale based on relevance, clarity, simplicity and ambiguity. The Content Validity Index (CVI) was used to measure the construct validity of the survey instrument based on these ratings. The items that had a CVI over .750 remained. Any CVI lower than .750 were to be deleted from the survey. Complete data sets of the judges' responses are located in Appendix G. The CVI for the items in Survey I ranged from .796 to 1.00. This indicates an overall moderately high construct validity.

Convergent and Divergent Validity. Both convergent validity and divergent validity were established by first comparing answers to another question measuring the same concept (convergent validity), then by measuring this answer to the participants' responses to a question that asks for the exact opposite answer (divergent validity). For

example, if a respondent answers “yes” to the question, “Do you think that there is a role for genetics and genomics in the practice of speech-language pathology?” (Item 28) then one may ask, “What knowledge, skills, and attitudes competencies should your students demonstrate upon graduation?” (Item 7) checking for convergent validity. This could then be followed by, “What areas of study does genetics and genomics play a role in a speech language pathology curriculum?” (Item 27), as a means to check responses for divergent validity.

Procedures for Data Coding

Coding the data involved the following; if the item had two responses, they were coded with either a 1 or a 2 (e.g., bachelor’s or master’s degree), or were coded with a 1 or a 0 (e.g., Do you hold certification?). Some items had four possible responses and/or involved Likert scales (e.g., 1-5 years; 6-10 years; 11- 20 years; 21+ years). For such items, the choices were coded as 1, 2, 3, and 4. Some items had more than four responses (e.g., in which State is your program located?) for which they were coded as 1, 2, 3, 4, 5 and so on. When an item presented with no response, it was coded as missing data and provided a 0.

Data Analysis

Data were exported from the database server (SurveyMonkey.com) into a Microsoft Excel file and the Excel file was imported into IBM SPSS version 19.0 (2010) for analysis. Data were examined using frequency counts, descriptive statistics (mean, standard deviation, median), as well as cross-tabulation (independent t-tests), and (where appropriate) Pearson Chi-square tests. The alpha was set at 0.05 for all tests of statistical significance. Survey results included categorical responses to the questions and Likert-type concept rankings, as well as textual comments. Demographic data were simply tallied for descriptive purposes. Textual data from comments or suggestions were collated and reviewed to identify common themes. (See Appendix I.)

Methodology and Research Design of Survey II: The Professional Speech-Language Pathologist

Subjects

The recruitment of respondents occurred through a "hybrid" approach; a combination of probability-based samples and convenience samples was used. A total of 1000 surveys were initially sent to randomly selected ASHA certified speech-language pathologists via email invitations. However, additional recruitment techniques were used when only 27 surveys were received in the first five days of recruitment. Subjects were 1,000 randomly selected ASHA-certified, professional speech-language pathologists in the United States. Two hundred thirty-two surveys of the randomly selected invitations were returned undeliverable. A response rate could not be computed as the respondents were recruited through a combination of approaches.

Methods

The data were gathered through the use of a large-scale, cross-sectional, census survey called *Genetics in the Post-Genomic Era: The Practitioners' Perspective*. The survey and accompanying cover letter (see Appendix I) were delivered by SurveyMonkey.com©, an online survey tool. Multiple methods of recruitment were utilized to reduce the possibility of low-response rates. Three methods were used to recruit participants for this study: (1) email addresses for potential respondents were obtained through databases; (2) the SurveyMonkey.com© link was posted on professional list servs and Internet-based professional and social networking groups (e.g., ASHA's Special Interest Groups (SIG) Online Community site – see Appendix J for a Special Interest Group Listing); (3) speech language pathologists were requested to forward the survey link to other members of their local professional network (snowball sampling). The potential responders were informed that their participation was voluntary with no incentive other than contributing to general knowledge.

As a means to facilitate a high response rate potential respondents were initially notified of the survey by a pre-survey, introductory letter (see Appendix K). The survey was subsequently sent to each prospective respondent three days upon receiving the notification.

The survey was sent to each prospective respondent three days after they received the introductory letter. One week after the initial mailing of the survey, non-responding speech-language pathologists were contacted and surveys re-sent. This was repeated twice. The surveys were collected from February 10, 2012 to March 11, 2012. Data entry for returned surveys, and statistical analysis, were conducted using IBM SPSS v.19.0 (SPSS, Inc, Chicago, IL) software package.

Survey Development

The approach to designing the survey instrument used for the professional speech-language pathologist was similar to the survey used for program directors of training programs described in this chapter (p. 51). See Table 7.

Defining the Content and Selection of Items

Procedures used in the content development are identical to those performed for Survey I (p. 52). The content domains were developed based on the questions raised from the literature review, benchmarks of genetic and genomic content identified by *Essential Core Competencies in Genetics for all Speech Language Pathologists* (NCHPEG, 2004, 2007), discipline specific concepts identified in the literature, and similar instruments used in medicine and allied health professions (Chermak & Wagner-Blitz, 1993; Lapham et al., 2000).

Five query domains (or concepts) were defined: (1) perceived attitudes of genetics and genomics in communication sciences of professional speech-language pathologists; (2) self-perceived knowledge of genetics and genomics; (3) qualitative genetic and genomic related services performed in clinical practice; (4) confidence levels; and

(5) qualitative, thematic concepts. Table 10 outlines the query domains and concepts mapped according to item number and source.

Query 1: Attitudes of Genetics and Genomics. Speech-language pathologists were asked about their awareness of recent genetic and genomic advancements (Item 1), their opinions of the impact of recent advancements in speech-language pathology (Item 2), and their opinions concerning how well their speech-language pathology training prepared clinicians to understand the current fields of genetics and genomics (Item 6).

The perceived knowledge of genetics and genomics as it relates to (1) basic science and theoretical principles, and (2) discipline-related genetics and genomics was assessed in Item 16. Respondents were asked to indicate their perceived level of knowledge of genetic and genomic topics, using a four-point Likert-type scale.

Query 2: Knowledge of Genetics and Genomics. The perceived knowledge of genetics and genomics as it relates to (1) basic science and theoretical principles, and (2) discipline related genetics and genomics was assessed in Item 16. The respondent was asked to indicate their perceived level of knowledge of genetic and genomic topics using a four-point Likert-type scale.

Query 3: Clinical Work Practices. Respondents were probed for their understanding of the areas within communication sciences and its disorders where genetics and/or genomics play a role (Item 8). The questions asked respondents to identify speech-language processes and disorders in which they perceived a role for genetics and genomics; whether they received questions from parents/families and patients about concepts in genetics and genomics, both in basic science terms (Item 9), and as these concepts relate to communication and/or disorders (Item 10); their general referral practices (Item 15); and the various clinical skills involved in the assessment, diagnosis, counseling and management of patients with communication disorders (Item 12). Area of practice (Item 26), and type of work setting (Item 27) were probed. Items 24 and 25 addressed current certification status and current engagement in clinical

Table 10 ***Mapping Survey Items to Domain, Concept and Evidence Constructs***

	Query Domains	Item	Concept Mapping	Source
	Professional characteristics	17, 20, 21, 22, 23	Types of graduate degrees offered, number of students	NA
	Educational characteristics	3, 4, 5	Presence, amount in genetic genomic science and applied content, delivery format (type, instructional methods).	Council on Academic Accreditation in Audiology and Speech Language Pathology (CAA).
1	Attitudes of genetics and genomics	1, 2, 6, 7, 8, 19, 20, 26, 27	Perceived importance and role of genetics and genomics	NA
2	Self-perceived knowledge of genetics and genomics related clinical services	16	Knowledge and skill of use of genetic and genomic principles as they relate to theoretical foundations (TF) and clinical applications (CA).	Core Genetic Principles in Genetics Essential for All Speech Language Pathologists (NCHPEG, 2007) Genetics Literacy Assessment Instrument for non-biology undergraduates (Vice-Bowling, Acra, Wang, Myers, Dean, et al., 2008) Statement paper, What does the speech-language pathologist or audiologist need to know about genetics when conducting assessments? (ASHA, 2006) Items developed de novo by the investigator.
3	Clinical work practices	8, 9, 10, 11, 12, 15, 24, 25, 26, 27, 28, 29, 30, 31, 32, 33	Current scope of practice	Core Genetic Principles in Genetics Essential for All Speech Language Pathologists (NCHPEG, 2007)
4	Self-perceived confidence levels	7, 11, 12, 14, 15, 24		N/A
5	Qualitative and thematic concepts	7, 16, 17		Genomic Education Rubric (Appendix G)

practice. Respondents were also asked to provide information about their teaching within the field of speech language pathology (Items 28 and 29). They were asked whether they are engaged in research that involves genetics and genomics (Item 30), and whether they had ever been or currently are a member of a craniofacial team or center (Item 31). For those indicating such team membership, additional questions requested information about location (item 32) and the role of the respondent as a member of the team or center (item 33).

Query 4: Confidence Levels. Finally, respondents were asked to indicate their perceived level of confidence in several areas: their understanding of current genetics and genomics as it applies to speech language pathology (Item 7); their ability to recognize a genetic and or genomic speech language related condition or disorder (Item 14); confidence in providing the appropriate information and counseling to families and patients (Items 11, 14); and confidence in performing various clinical activities (Item 12). Additional items asked where the respondent would refer, if a suspected genetic and/or genomic issue was identified (Item 15), and probed the respondents' level of confidence in understanding genetics and genomics in recent advances within the role of speech language pathology (Item 34).

Query 5: Determining Thematic Concepts. Thematic analysis of items 8 and 16 were mapped to four cognitive and pedagogic dimensions of the rubric described on page 55.

Survey Structure and Format

The survey for professional speech-language pathologists followed a similar structure and format to that used in the survey for program directors. The initial questionnaire began with a pool of 40 items aligned across the query domains. All questions were created *de novo*, and underwent several modifications. The question formats were the same as those used in Survey 1: ratings (e.g., Likert scales), split-

folding questions, and inverted funneling questions. The survey instrument included both closed-ended and open-ended questions. Answers were not forced, so that the survey could be advanced if a question was unanswered. Several questions were formatted with skip logic, subsequently varying the sample size of the items. Free text areas at the end of each question solicited comments or suggestions about additional important concepts in that category.

Bias Reduction

Methods used to reduce response bias in Survey II for speech-language pathologists were identical to those methods used in Survey I for program directors (p. 58).

Establishing Survey Reliability and Validity

Representational Sample. Portions of the sample were randomly selected, however participants were also solicited through snowball sampling and through professional community sites. Therefore, the sample was not random, and the non-random sampling was validated by comparing whether selected characteristics of the population of interest (215 speech-language pathologists) were represented in the sample.

Reliability. The data primarily represented census information, and not concepts upon which reliability analysis can be performed. However, when appropriate, a Cronbach's Alpha test was performed when an item represented a constructional theme across several items. For example, Items 7, 11, 13, and 14 represented confidence constructs. Cronbach's Alpha was performed. An alpha of .808 was achieved, indicating moderately high internal consistency among the scales.

Validity.

Content Validity, The methods used to evaluate content validity for the survey were similar to the theoretical constructs methods used to establish content validity for

Survey I. Content validity was established by a panel of judges within a focus group and pilot study.

Focus Group (Cognitive Interviews)

The focus group procedure for Survey II was similar to that used with Survey I. A panel of four judges (one geneticist, and three certified speech-language pathologists) participated in the focus group. The reviewers' feedback was used in the selection of the items for the final version and for revision of items; thus the content validity of the instrument is supported by the reviewers' participation in its development. See Appendix L for responses of focus group. See p. 64 for details on the procedures used.

Based on the results of focus group interviews, six items were deleted from the original pool of 40 items due to excessive length, redundancy of concepts, and ambiguity in regards to question format. Items indicating ambiguity, overlapping of questions, poorly constructed questions, and redundancy were revised. On the basis of the reviewer's responses to the questions and their individual comments, the items once again underwent revision. This resulted in the final 34-item survey.

Pretesting of Survey – Pilot Study

The survey was administered to eight ASHA certified speech-language pathologists randomly selected from the member directory of the Westchester Speech-Language Hearing Association. Similar procedures were followed as for the program directors' pretest, outlined on page 60. The CVI for the items in Survey II ranged from .786 to 1.00, indicating an overall moderately high content validity (see Appendix M).

Data Analysis and Data Coding

Methods of data analysis and coding for Survey II were identical to those described for Survey I (p. 61).

Summary

To summarize, two surveys were developed to characterize genetic and genomic education in speech-language pathology. The first survey was designed for program directors of academic training programs in the field, and the second was designed for practicing speech-language pathologist. Both surveys underwent iterative evaluation procedures to ensure internal consistency and content validity. Survey I consisted of a 32-item instrument designed to characterize program directors perceptions of genomic in the education of students in a speech –language pathology degree program. A population of 242 program directors was invited to participate in the study. Survey I was fully completed by 65 program directors, resulting in a 28% response rate. Survey II, a 34-item tool was structured to characterize professional speech-language pathologist perceptions of genomics in education and clinical practice. Seven hundred sixty-one random and non-randomly selected certified speech-language pathologists were invited to participate in the survey. Two hundred and fifteen completed surveys were returned, representing a 35% response rate. Results of both surveys are presented in Chapter IV.

Chapter IV

RESULTS

Degree Programs in Speech-Language Pathology

The presentation of the data analysis, as previously described in Chapter III, is organized by the two groups surveyed: program coordinators of degree programs in speech-language pathology, and professional speech-language pathologists. The first objective of the present study was to characterize the current state of genomic education in the training of speech-language pathologists. Results of the Survey I called *Genetic Content of Speech Language Pathology Programs* are presented as per the three queries outlined in Chapter I (p. 1). The queries are mapped to the objectives of the study: (1) Characterize program directors' perceptions of genetics and genomics; (2) Identify and characterize genetic content and extent of delivery format in degree program curricula; and (3) Characterize the thematic trends of the perceptions of the program directors. Each query result will conclude with a summary. The survey questions within each query are included. Results of Survey II will follow.

Survey Response Rate

Data from the online survey, SurveyMonkey.com© were imported into IBM SPSS version 19 (2010) for analysis. Of the e-mail invitations sent to 242 United States programs directors of degree programs in speech language pathology, four invitations was undeliverable (three opted out; one bounced), resulting in 238 distributed invitations. When the survey was closed four weeks later, 75 degree programs responded to the

survey. However, seven surveys were not fully completed, and were thus not included in the analysis, resulting in a total of 68 completed surveys. This represented a 28% response rate. Not all participants responded to every question, therefore, the sample size for individual items varied across the survey.

Program Director Characteristics

Question 30. Are you the program director of the speech-language degree program?

Table 11 summarizes the demographic characteristics of the program directors. Ninety-four percent (60/64) of the survey responses were filled out by the program directors. Faculty members other than the program director filled out four of the returned surveys.

Table 11. ***Program Director Characteristics***

Program Director Characteristics*	Total Number of Program Directors Responding to Question	Frequency of Responses	Percent
Program directors completed survey	64	60	94%
Faculty member completed survey		4	6%
Years in current position	63		
0-5		14	22%
6-10		13	21%
11-15		10	16%
16-20		8	13%
.21+		18	29%
Area of training	63		
Speech Language Pathology		51	81%
Dual Speech-Language Pathology/Audiology		4	6%
Speech Scientist		4	6%
Audiology		3	5%
Hearing Scientist		1	2%

*National population demographics were not available for this variable.

Question 32. In what area are you trained?

Eighty-one percent (51) of the program directors indicated they were trained as speech-language pathologists. Four respondents reported they were dual certified in both speech-language pathology and audiology. Table 12 shows the responses per each area of training.

Table 12. ***Area of Training of Program Directors***

Area of Training of Responding Program Directors N=63	Frequency	Percent
Speech-Language Pathologist	51	81%
Speech Scientist	4	6%
Dual Certified Audiologist and Speech-Language Pathologist	4	6%
Audiologist	3	5%
Hearing Scientist	1	2%

Question 29. How long have you held your position in the speech-language pathology department?

Twenty-nine percent (18) of program directors held their respective position for twenty years or more, while twenty-two percent (14) of program directors reported being in their position for less than five years. Eighty-one percent (51) of program directors were ASHA certified speech-language pathologists. (See Table 12.)

Degree Program Characteristics

Question 3. How long has your institution been offering a program in speech-language pathology?

Eighty-one percent (54/67) of programs have offered a degree program in speech-language pathology for 21 years or more. This was followed by 15% (10) of programs having offered a program for 10 to 20 years, and with five programs having offered a speech-language degree for nine years or less (see Table 13).

Table 13. ***Number of Years Offering Program in Speech-Language Pathology***

Number of Years Offering Degree Program (N=67)	Frequency of Response by Program Directors	Percent
20+ years	54	81%
16-20 years	4	6%
11-15 years	3	5%
6-10 years	3	4%
0-5 years	2	3%

Question 4. What degree(s) does your program offer in speech-language pathology?

The types of degrees offered by the training programs (69) are listed in Table 14. Fifty-five percent (37) of the programs offered a Bachelor's and Masters degrees, while twenty-one percent (14) offered a Masters degree only. Twenty-one percent of the programs (14) offered all three: Bachelors, Masters, and Doctorate degrees, and two programs offered a Bachelor's degree only.

Table 14. ***Type of Degree Offered In Speech-Language Pathology Programs***

Types of Degrees Offered in Responding Speech-Language Pathology Degree Programs	National Population* N=235	Percent	Frequency of Survey Response n=67	Percent
Bachelor and Masters	140	60%	37	53%
Bachelor, Masters and Doctorate	56	24%	14	20%
Masters Only	16	6%	13	19%
Masters and Doctorate	7	2%	2	3%
Bachelor	2	1%	1	1%
Other Speech-Language Pathology Degree	11	5%	0	0%
Other Doctorate	3	1%	0	0%

*www.asha.org/edfind/results.aspx?area=SLP&degree=Masters+locationAll
www.asha.or/students/academic/graduates/-school.

Programs Located by Geographical Region

Question 2. In what state is your program located? All four geographical regions of the contiguous United States (US Census Bureau, 2010) were represented by the 65 program directors that responded to this question. The geographic region of the Northeast, with 34% (23) was the most represented. Regions of the South and Midwest were equally represented with 21% (14) of program directors responding to the survey. Eight of the program directors represented the Southwest and six of the program directors representing degree programs in the West (see Table 15). Appendix N provides the each of the states per geographical region.

Programs in the following states were not represented; Hawaii, Kentucky, Louisiana, Maine, Maryland, Michigan, Minnesota, Montana, Nebraska, Nevada, North Dakota, Ohio, Oregon, South Carolina, South Dakota, Utah, Virginia, Washington, West Virginia, and Wyoming. The states of Alaska and Delaware do not offer a degree program in speech-language pathology.

Table 15. ***Geographical Areas of Responding Program Directors***

Location of Degree Program by Geographical Area	National Population of Degree Programs per Region	Percent	Frequency of Response of Program Directors	Percent
Number of degree program in the US	242			
Number of responding program directors			65	
Northeast	55	23%	23	34%
South	86	36%	22	34%
Midwest	67	28%	14	21%
West	34	14%	6	9%

Medical Affiliations

Question 5. Is your speech-language pathology program affiliated with a medical school or teaching hospital? Programs in which there exists a relationship with a medical school and or teaching hospital may provide their students and faculty access to specialists in the field of genetics and genomics, and the specialists may have a greater presence in the pedagogy of the curriculum. Of the sixty-eight program directors that responded to this question, 12 program directors (18%) indicated they do have an affiliation with a medical school and or medical teaching facility.

Question 6: Is your speech-language pathology program designated as a medically-based? A medically-based training program is one which provides increased medical emphasis in speech language pathology training, specifically to those interested in a professional career working in hospitals or related facilities. Of the 67 programs represented, seven program directors (10%) indicated their programs were designated as medically based.

Results were analyzed further to reveal any association between being a medically affiliated and a medically-based program. Results of a Pearson Chi-square analysis indicated a significant association (χ^2 (1), =19.30, p< .001). Each of the 12 programs that were reported to be medically affiliated with a medical school was also likely to be also designated as medically-based.

Degree Programs Engaged in Genetic Research

Question 22. Does your department engage in genetically-based speech or language research? Program directors were asked whether their program engaged in genetic and or genomic related research. Six programs reported nine research projects being conducted at the time of the survey. All reported research was being conducted at the doctoral level.

Question 23. What area(s) of research is currently being conducted? The areas of reported research varied and are listed in Table 16. The research topics reported by the

program directors were classified into whether the topic reflected research in genetics (single gene disorders) or whether the topic represented research in genomics.

The genetics classification was defined as the study of primarily Mendelian inheritance and single gene disorders related to structural anomalies (e.g., Trisomy 21, Down syndrome). Genomics in this context was defined as the study of polygenic and mulitfactorial inheritance (combination of environmental, genetic and stochastic factors). The following are examples of polygenic and multifactorial inheritance disorders; autism, apraxia, diabetes, and schizophrenia. Results of the classification indicate that four of the research topics reported by the program directors appear to represent genetics and three research topics represented a genomics model. Two of the topics, "Fluency" and "Many- adult neurogenic language, cognition, dysphagia, and voice" were not well described by the program director to adequately determine which model they represented.

Table 16. ***Topic Areas of Genetic and Genomic Research Being Conducted in Degree Programs***

Topics Reported Genetic and or Genomic Research Being Conducted in Degree Programs n=9	Number Research Projects	Genetic Research Topics	Genomic Research Topics
"Craniofacial abnormalities"	2	+ +	-
"Cleft Palate and Huntington Disease"	1	+	-
"Dysphonia secondary to Inherited Elastin gene (ELN) abnormalities"	1	+	-
"Neurodevelopmental syndromes"	1	-	+
"Specific Language Impairment and Developmental Dyslexia"	1	-	+
"Speech Sound Disorders"	1	-	+
"Fluency"	1	Undetermined	Undetermined
"Many- adult neurogenic language, cognition, dysphagia, and voice"	1	Undetermined	Undetermined
Total	9	4/6	3/6

Query 1. Program Directors' Perceptions and Attitudes of Genetics and Genomics

Inquiring as to the perceptions and attitudes of program directors provides a characterization of the "environment" of the program. It provides awareness of current genetics and genomics, and how this may influence or guide the program's faculty and or student body in regards to interest in genomic curricular development and or research. Several variables demonstrating perceptions and attitudes of genomic education were mapped across the survey; perceived interest in genetics and genomics, perceived role and impact of genetics and genomics, level of graduate preparedness, and expected competencies of graduated students.

Interest in Genetics and Genomics.

Question 20: In your opinion, how much interest in genetics/genomics is expressed by your students? As a means to determine if and to what extent program director's were aware of the expressed interest in genetics and genomics amongst the student body and faculty members of their degree program, they were asked to respond to a 5-point Likert-scale (1- *no interest*; 2- *some interest*; 3- *moderate interest*; 4-*significant interest*; and 5- *don't know*). Table 17 presents the frequency and percentage of responses. Forty-eight percent (31/64) of their students expressed "*some interest"* in genetics and genomics, followed by 39% (25) who responded "*moderate interest,"* four reported *"significant interest"*, and one program director indicated their students expressed *"no interest"*.

Question 22: In your opinion, how much interest in genetics/genomics is expressed by your faculty? In regard to faculty and their expressed "interest" in genetics and genomics, 52% (34/65) program directors responded their faculty expressed "*some interest"* in genetics and genomics, followed by 26% (17) reporting "*moderate interest,"* eight responding program directors expressed *"significant interest,"* and four of responding program directors indicated their faculty expressed *"no interest."* See Table 17. Three program directors indicated they *"did not know"* the expressed interest

of their students. Two program directors indicated they did not know the expressed interest of their faculty.

Table 17. ***Frequency and Percent of Program Directors Extent of Interest Expressed by Students and Faculty***

Program directors perceived level of interest in genetics and genomics N=65	Program directors perceived student interest n=64		Program directors perceived faculty interest N=65	
	Frequency of Response	Percentage	Frequency of Response	Percentage
No interest	1	2%	4	6%
Some interest	31	48%	34	52%
Moderate interest	25	39%	17	26%
Significant interest	4	6%	8	12%
Don't know	3	5%	2	3%

Role and Impact of Genetics and Genomics.

Question 25: In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology? Perceptions regarding the impact and role of genetics and genomics in the field of speech-language pathology often reflects a level of awareness of current advancements and an increased understanding of the current role the genetic and genomic advancements may have in the field of speech-language pathology. Of the sixty-five program directors responding to an inquiry as to their perception of the impact current genetics and genomics on speech-language pathology, 40% (26/65) of program directors felt that genetics and genomics had a "significant impact" on the field of speech-language pathology, followed by 34% (22) who felt a "moderate impact," and with 15 of the responding program directors reporting a "minimal impact. (See Table 18.)

Table 18. ***Program Director's Perception of Impact and Perceived Future Change on Impact***

	Perceived impact of current genetic and genomic advancements on speech-language pathology		Perceived future change on impact of current genetic and genomic advancements on speech-language pathology	
Level of impact	N=65	Percentage	N=65	Percentage
Significant impact	26	40	13	20
Moderate impact	22	34	36	55
Minimal impact	15	23	17	26
No impact	0	0	0	0

Question 27: Do you believe genetics/genomics has a role in the clinical practice of speech-language pathology? When asked their opinion of the role genetics and genomics play in the *clinical practices* of speech-language pathologists, 95% (62/67) of program directors indicated "Yes", genetics and genomics does "*play a role*" in the clinical practice of speech-language pathologists. Three program directors reported they did not believe genetics and genomics played a role in clinical practice.

Question 28: Of the following areas of study in a speech-language pathology curriculum, which one(s) do you believe genetics/genomics has a role? Further elaboration of the role of genetics and genomics was requested when program directors were asked to rate via a 4-point Likert-scale (1-no role; 2-some role; 3-moderate role; and 4-significant role) the role that genetics and genomics may play in typical areas of study included in the curriculum (e.g., areas may include courses in anatomy, sound disorders, and language delay/disorders, etc.). A list of all 21 areas of study and the program directors response, ranked per frequency and percent is located in Appendix O. An abbreviated summary is presented in Table 19 with the three most frequent responses provided. Sixty-one percent (36/59) of the program directors indicated that genetics and genomics played "a significant role" in the area of *hearing.* This was followed by the

area of *adult language* having “some role” by 59% (33) of the program directors, and *reading*

Table 19. ***Program Director's Perceived Role in Various Areas of Study (Abbreviated)***

Program Directors Perceived Role of Genetics and Genomics in Areas of Study N=60														
No Role	*f*	%	**Some Role**	*f*	%	**Moderate**	*f*	%	**Significant**	*f*	%	**Don't Know**	*f*	%
Phonetic Acoustics	19	37	Adult Language	33	59	Reading	25	41	Hearing	36	61	Speech Science	3	5
Speech Science	13	24	Swallowing	29	50	Anatomy	24	41	Autism	28	47	Swallowing	3	5
Adult Articulation	9	16	Adult Articulation	18	48	Pediatric Language	24	41	Resonance	21	38	Physical Assess	3	5

having “a moderate role” by 41% (25) of the responding program directors. Of interest, 37% of the program directors indicated that genetics and genomics “did not play a role” in *phonetic acoustics*. Twenty-four percent of the responders reported that areas of *speech science,* and nine of the program directors reported the area *adult articulation* as not having genetics and genomics role. A number of program directors indicated they “did not know” the role genetics and genomics played in the presented areas of study. For example, 63% of respondents indicated they did not know the role genetics and genomics played in the area of *speech science.*

Question 26: With continued advances in genetics/genomics, do you think the impact on clinical practices of the speech-language pathologist will change in the next 5-10 years? Fifty-six percent (36/65) of program directors indicated that with continued advances in genetics and genomics, the clinical practices of speech-language pathology would “*moderately change”* in the next 5-10 years from its current level. This was contrasted to 26% (17) of directors who felt a *“minimal change”* and 20% (13) of the program directors who responded that “*significant change”* would occur.

Expected Genomic Competencies of Graduating Students.

Question 7: The following is a list of knowledge, skills, and attitudes often required by a graduated speech-language pathologist. Please check all competencies your speech-language program expects your students should demonstrate upon graduation. Graduated students are expected to demonstrate a level of knowledge (literacy and competency) in various subject areas across constructs of knowledge, skills and attitudes. The construct of attitude will not be used as a research variable in this investigation. Although attitude is an integral part of overall literacy and competency of professional education, it is felt to be an attribute developed over time and with experience, which students had not had the opportunity to develop in the two years of a training program. Therefore the current research will focus on the knowledge attributes of literacy and competency.

Genetic and genomic literacy includes the underpinnings of theoretical concepts of various constructs mapped across varied learning opportunities. Training programs in speech-language pathology typically provide opportunities for the development of tacit knowledge or competency, gained as a result of experience in performing certain tasks within onsite and external practicum opportunities.

To understand the program director's perception of what type and level of genetic and genomic literacy and competency constructs are required of their students program directors were asked to respond to several questions mapped to the attributes of literacy and competency. These concepts were based on the "knowledge" and "skill" competencies outlined by NCHPEG, (2007) in the Essential Genetic Core Competencies for all Speech Language Pathologists.

Program directors were asked to indicate from an 18-item concept list, which constructs of expected behaviors of genetic and genomic literacy and competency were expected from their graduating students. Multiple responses were accepted. Table 20 specifies program directors responses to expected constructs and the frequency in number and percentage. Results indicated that of the 68 responding program directors, 96% indicated that genetic and genomic related constructs were expected of graduating students. Only one respondent indicated that the competency constructs offered were not expected by graduating students.

Types of Competencies Expected. The program directors perceived "competencies" were descriptively analyzed to determine the possible underlying conceptual understanding of each presented construct. Table 20 depicts the results with regard to ranked frequency and percent. Over 88% (59/68) of the program directors indicated that "Knowledge of craniofacial development/embryology" construct was an expected competency of a graduating student.

Table 20. ***The Frequency and Percent of Competencies Expected from Graduated Students***

Literacy and Competency Constructs as per NCHPEG (2007)	Frequency of Program Directors' Response N=68	Percent
1. Knowledge of craniofacial development/embryology	59	88%
2. Recognize when a speech language and or hearing issue is potentially related	58	87%
3. Recognize common genetic syndromes	58	87%
4. Make referrals for genetic assessment and counseling	50	75%
5. Recognize at risk family members based on family history	44	66%
6. Make appropriate genetic referrals based on diagnostic information	36	54%
7. Knowledge of patterns of inheritance	35	52%
8. Correct misconceptions about genetic and or environmental causation of speech, language or hearing disorders and delays	34	51%
9. Counsel on impact of condition on family	33	49%
10. Knowledge of dysmorphology	32	48%
11. Discuss the genetic nature of a speech language delay or disorder	32	48%
12. Make prognostic judgments of susceptibility risks of occurrence or recurrence of communication disorders	31	46%
13. Assess the genetic contribution to a speech language hearing diagnosis	26	39%
14. Obtain a genetic history in relationship to a communication disorder or delay	21	31%
15. Discuss genetic causation with individual and or family member	16	24%
16. Counsel on legal, ethical and social issues regarding a genetic related diagnosis and or condition	5	7%
17. Counsel on various types of genetic tests available for assistance in diagnosis and etiology	4	6%
18. Knowledge of molecular pathogenesis	2	3%
19. None of these	1	1%

This was followed by construct, "*Recognize when a speech language and or hearing issue is potentially related"* and *"Recognize common genetic syndromes"*, each

ranked by 87% (58) of the program directors to be an expected competency. Each concept in this item was categorized (+/- value) as to whether the concept represented "genetics" versus "genomics". In fact, nearly three quarters of the competencies reported by the program directors were related to genetics (Items 1-7, and 10). Few program directors expected genomic related competencies from their graduated students (Items 8, 9, 11-17).

The concepts were further categorized (+/- values) as to whether they represented competencies involved in theoretical knowledge, assessment/diagnosis, and management/counseling. Results indicated four concepts were related to theoretical knowledge, five concepts were related to assessment/diagnosis, and nine concepts were related to management/counseling. Competencies concerning management/counseling were reported by more program directors (241) than competencies involving assessment/diagnosis (207).

Preparedness of Graduated Student.

Question 24: In your opinion, do you believe the graduated speech-language pathology student is prepared with the appropriate knowledge and skills in genetics/genomics? Program directors were asked whether their graduated students were prepared with the appropriate knowledge and skills in genetics and genomics. Thirty-six percent (32/64) of the responding program directors reported "Yes," and 50 % reported, "No" they felt their graduated student was not prepared. Nine responding program directors reported they did not know.

Query 1. Summary. Overall, program director's perception of the impact and role of current genetic and genomic advancements reflected a supportive environment for the program's student body and faculty. Program directors reported their students express more interest in genetics and genomics than their faculty. Half of the responding program directors felt their graduated students were not prepared in the knowledge and skills in genetic and genomics. Practically, all program directors expected genetic and genomic

related competencies from their graduated students with those competencies related to genetics as being the most frequent expected competencies by program directors. Competencies involving management/counseling were reported by more program directors than competencies of assessment/diagnosis.

Query 2. Presence and Extent of Genetic and Genomic Content

Query 2 inquires of the presence, and delivery format of genetic and genomic content in the curricula of speech language pathology training programs. The following results were obtained.

Question 8. Does your speech-language pathology program include genetic/genomic content in your speech-language pathology curriculum?

No Genetic and/or Genomic Content Offered. Twenty-eight percent (19/48) of the program directors indicated their curriculum does not offer genetic and or genomic content.

Question 9. What may be the reason(s) why genetic/genomic content is NOT being offered? These program directors were asked to provide possible reasons why genetic and genomic content was not offered in their curriculum. Multiple responses were accepted and are reported in Table 21. Of those who provided responses, the reason(s) as to why a curriculum with genetics content was not being provided were overwhelmingly related to lack of trained and qualified faculty (89%). This was followed by concerns of added workload for students (74%), and increased workload of faculty (58%). The rationale, *genetic and genomic content is not an accreditation requirement* was reported by eight responding program directors as a reason for not providing genetic and genomic content.

Question 10. Are there plans to develop genetic and genomic content in the curriculum within the next five years? Five of the program directors that did not currently provide genetic and genomic content, indicated they were planning on

developing a genetic and genomic content in their program's curriculum within the next five years. Thirteen of the programs that do not offer genetic and/or genomic content in their curricula indicated they do not plan on developing a genetic and genomic content within the next five years.

Table 21. ***Reasons for Lack of Genetic and Genomic Content in Degree Programs***

Reasons Given for Not Providing Genetic and Genomic Content	Frequency of Responses N=19	Percent
Genetics/genomics is relevant to the communication sciences, yet do not have trained personnel	17	89%
The addition of genetic/genomic course and or content would provide increased workload for students to an overcrowded curriculum.	14	74%
The addition of genetic/genomic course and or content would provide increased workload application of additional teaching commitments	11	58%
It is not a requirement of the accreditation criteria	8	42%
Providing genetic/genomic course and or content is cost prohibitive	4	21%
Do not know what genetic/genomic course and or content is relevant to speech language pathology	1	5%
Genetics/genomics is not relevant to the training of speech-language pathologist	0	0.0%
There is a lack of interest in the department to provide genetic/genomic content in the curriculum	0	0.0%

Programs Offering Genetic and/or Genomic Content

Seventy-two percent (47/67) of responding program directors indicated their programs offered genetic and or genomic content in their curricula.

Genetic Genomic Content Format.

Question 11. Describe the structure of the genetic/genomic content offered in the curriculum. Provide the percent of genetic/genomic content. Program directors were asked to describe the format in which the genetic and genomic content was presented. Generally, four models of delivery were reported: (1) stand-alone course with the

primary topic being science-based genetics and genomics only, (2) stand-alone course with the primary topic being genetics and or genomics of speech language pathology (or communication sciences), (3) genetic and genomic content integrated into a craniofacial disorders course, and (4) content integrated across several or all courses offered in the curriculum.

Fifty-seven percent (27) of the responding program directors reported offering a genetic and genomic content integrated within a course designated for craniofacial disorders. Twenty-five percent (12) of program directors reported offering genetics and genomics integrated across pre-existing speech-language pathology courses. Seventeen percent reported offering a stand-alone course specifically oriented to the genetics of speech-language pathology, and a stand-alone genetics and or genomics course(s) was reported to be offered in one degree program. (See Table 22.)

Of those programs that offered genetic and genomic content in a pre-existing course, directors were asked to provide an estimate of the percent of genetic and genomic content included. Table 23 provides the reported percent of genetic and genomic content. Thirty-three percent of directors reported that less than 10% of the content was designated genetic and or genomic.

Table 22 ***Type of Delivery Format of Genetic and Genomic Content***

Delivery Format of Genetic and Genomic Content	Frequency of Response n=48	Percent
Integrated content into craniofacial course	27	57%
Integrated content into existing speech-language pathology course	12	25%
Stand-alone genetics and speech-language pathology	8	17%
Stand-alone genetics only	1	2%

Table 23 ***Percent of Genetic and Genomic Content in Pre-existing Courses***

Percent of Genetic and Genomic Content in Pre-existing Courses	Frequency of Response n=12	Percent
0-10%	5	33%
11-20%	4	25%
21-30%	1	11%
31-40%	1	1%
41-50%	1	1%
51-60%	0	0%
61-70%	0	0%
71-80%	0	0%
81-90%	0	0%
91-100%	0	0%

Method of Content Delivery.

Question 12. How often is the course offered?

Question 19. What type of instructional method is typically used in teaching the genetics/genomics course(s)?

Ninety-six percent (45/47) of the program directors reported the method of content delivery was primarily didactic teaching (classroom) accompanied by use of small group discussions (36%), guest seminars (25%), nine responding program directors reported web-based instruction integrated with classroom lectures, and six responders reported distance education instruction. One program director reported using a web-based only method of presenting genetic and genomic content into the curriculum, and one program director reported they “didn’t know.” No one reported genetic and genomic content delivered through contexts where students could apply theoretical concepts (e.g., working onsite with individuals with communication disorders) to clinical settings.

Question 13. Is this a required or an elective course?

Question 14. If it is an elective course, approximately what percentage of students are enrolled in the class each time it is offered?

Question 15. In what year of the student's degree program is a course(s) with genetic/genomic content typically taken?

Seventy-eight percent (32/40) of the courses which offered genetic and genomic content were considered as a program requirement (as compared to an elective course). The course which included genetic and genomic content was reported to be offered once a year by 75% (30) of the programs, once every semester by three of the programs, and once every other year by three of the degree programs. If a stand-alone course was offered as an elective, the class size ranged from ten to 50 students. The genetics and or genomics courses were offered to students in their second year of study by 44% (19) of the degree programs, while 35% (15) of the programs offered genetic and genomic content integrated into the curriculum throughout the student's training (across all years).

Instructors.

Question 18. Describe the characteristics of the instructor teaching the course with genetic/genomic content. Ninety-eight percent (44/45) of the instructors were speech-language pathologists. Six of program directors indicated that the instructor had conducted research in genetics but did not have a degree in genetics and or genomics. One program director responded that a genetic counselor was the instructor, and one instructor had a degree in biology.

Courses with Integrated Genetic and Genomic Content.

Question 16. In which course(s) is genetic/genomic content integrated? The course(s) offered in which genetic and genomic content were integrated into pre-existing courses within the curriculum is presented in Table 24. Eighty-four percent (38/45) of the program directors reported genetic and genomic content was included in a Craniofacial Disorders/Disabilities course. Sixty-four percent (29) reported integrating content into

courses of Disorders of Speech; sixty-four percent reported integrated content in Disorders of Language, and courses integrating content into courses involving Hearing were reported by 38% of the program directors to integrate genetic and genomic content. See Table 24 for all courses listed with genetic and genomic content.

Table 24. ***Courses Reported by Program Directors with Integrated Genetic and Genomic Content***

Speech Language Pathology Course Type in which Genetic and Genomic Content is Integrated	Frequency of Responses n=46	Percent
Craniofacial disorders/disabilities	38	84%
Disorders of speech	29	64%
Disorders of language	29	64%
Hearing	24	53%
Neuroscience	17	38%
Diagnostic methods	16	36%
Anatomy	13	29%
Dysphagia	12	27%
Disorders of cognition	12	27%
Articulatory physiology	9	20%
Management	6	13%
Professional issues	5	11%
School-based services	3	7%

Genetic and Genomic Topics Integrated into Curriculum.

Question 17. What topics of genetics/genomics are covered in your curriculum?

Program directors were asked to select from a list of genetic and genomic topics which, in their opinion are included in their genetic and genomic curriculum. The topics were concepts representing basic genetic science principles and discipline specific genomic principles based on the Core Competencies (NCHPEG, 2004, 2007). The topics were mapped to knowledge (literacy and competency) constructs involved in the

assessment/diagnosis and management/counseling of individuals with communication disorders.

Ninety-six percent of the program directors (42/44) indicated “communication disorders specifically related to genetics” as a topic included in the genetic and genomic content. This was followed by 88% (39) program directors indicating “characteristics of genetic conditions/ syndromes,” while 86% (38) indicated “importance of family history” being a topic included in the genetic and genomic content. All topics reported to be included in the genetic and genomic content is depicted in Table 25.

Table 25. ***Topics Integrated into Genetic and Genomic Content***

Genetic and Genomic Topic	Frequency of Response n=45	Percent
Specific genetically-related communication disorders	42	96%
Characteristics of genetic conditions/syndromes	39	88%
Importance of family history	38	86%
Craniofacial development/embryology	37	84%
Patterns of inheritance	33	75%
Making referrals	29	66%
Overview of genetics in communication sciences	27	61%
Indications for genetic testing	20	45%
Embryology	18	41%
Prognosis of genetic disorders/diseases	14	32%
Recurrence risks	12	27%
Genetic counseling	12	27%
Treatments for genetic disorders	12	27%
Legal, social and ethical issues	10	23%
Cell biology	9	20%
How to perform a pedigree analysis	6	14%
Epigenetics	6	14%

Query 2 Summary. Seventy-three percent of program directors reported offering genetic content in their speech-language pathology curriculum. The format is primarily integrated into pre-existing courses, namely a craniofacial course, held usually once a year. The instructors were overwhelmingly speech-language pathologists with no formal training in genetics and or genomics. Sixty percent to eighty percent of the program directors reported including genetic and genomic content in a course of craniofacial disorders, disorders of speech, disorders of language and hearing disorders. The genetic and genomic topics typically integrated in the courses were primarily related to genetics and single gene disorders as compared to genomics. The lack of qualified, trained instructors was the primary reason given as to why genetic and genomic content was not offered.

Query 3: Thematic Analysis of Genetic and Genomic Concepts in Curriculum

Thematic analyses were performed to investigate the presence of trends in the data. Group means, standard deviation, and paired t-tests were performed on the four quantifiable dimensions of the Genomic Education Rubric; genetic versus genomic approach (*Genetic Genomic Dimension*), use of basic genetic sciences versus discipline specific information (*Content Dimension)*, type of knowledge expected; literacy versus competency (*Knowledge Dimension*), and type of pedagogic approach; whether awareness versus conceptual constructs (*Pedagogic Dimension*) are used in training programs where genetic and genomic content is offered.

The dimensions were mapped across program director's responses to; Item 7 (Genetic Genomic Competencies Expected of Graduating Students), Item 16 (Courses in Which Genetic and Genomic Content are Integrated), and Item 17 (Genetic and Genomic Topics Covered within Genetic and Genomic Curriculum). See Appendix E for full description of the rubric used. The results of the thematic analysis for each dimension follows.

Genetic/Genomic Dimension. The year 2003 represents the emergence of a new way of thinking about genetics, as introduced in Chapter I. Prior to the completion of the Human Genome Project, "genetics" represented the field. Since 2003, the field of genetics has been divided into two approaches; *genetics,* the study of single genes and Mendelian modes of inheritance, and *genomics* the study of multiple genes and multifactorial modes of inheritance (including epigenetics). Results are reported in Table 26.

Results of the thematic analysis were that on average, programs offering genetic and genomic content expected more genetic based competencies (*M*=5.37; *SD*=2.58) as compared to genomic based competencies (*M*=2.36; *SD*=1.28), *t*(66)=12.09, *p* <.000.

Table 26. ***Thematic Analysis of Programs Offering Genetic and Genomic Content***

Survey Item	Rubric Themes Type of Model	Rubric Dimensions	Frequency of Response N=67	Mean	*SD*	*t*	*df*	*Sig.*
Q 7, 16,17	Genetic and genomic model	Pre-genomic	67	5.37	2.58	12.10	66	.000
		Post genomic	67	5.49	1.28			
Q7, 16, 17	Content model	General genetic genomic knowledge	67	5.49	2.72	15.11	66	.000
		Discipline specific knowledge	67	2.96	1.48			
Q7, 16	Knowledge model	Level of literacy expected	67	6.70	2.49	22.13	66	.000
		Level of competency expected	67	1.85	1.28			
Q16	Pedagogic model	Conceptual knowledge	67	8.12	3.15	20.71	66	.000
		Process knowledge	67	.55	.502			

Content Dimension. Content dimension assesses the type of concepts used in the curriculum, whether the curriculum was directed towards basic genetic science content (e.g., analysis of Punnett's square in inheritance), or whether content was focused on discipline specific topics (e.g., the type and number of genes related to stuttering and its relevancy to etiology and assessment of the disorder).

Competencies with basic *genetic science* content (M=5.49; SD=2.72) were on average, rated with higher frequency by program directors as compared more *discipline specific competencies* (M=2.96; SD=1.48, $t(66)$=15.11, p<.000. The content of the genetic oriented curriculum tend to be directed at basic genetic science as compared to genomics that was directly related to speech language pathology.

Knowledge Dimension. An opportunity to gain a theoretical foundation or a degree of literacy is integral to the education of speech-language pathologists. Equally integral is the provision of the opportunity to "exercise" theoretical concepts within contexts of clinical application. In addition to the classroom setting, each training program offers the student ways to gain experience and training (competency) working onsite with faculty providing supervision.

Results indicated that responding program directors on average, rated with more frequency, competencies and courses which provided more of a theoretical approach (M=7.36; SD=2.12) when compared to competencies and courses where skills in clinical application or competency was represented (M=5.26; SD=2.58, t(66)=22.13, p<.000).

Pedagogic Dimension. How the genetic and genomic content is shared with students was of interest. The pedagogy used in a genetic and genomic curriculum frames how and what is expected from the student in regards to knowledge. The theme of inquiry, whether the genetic and genomic content, was delivered in regard to conceptual (awareness knowledge) approach versus a process approach. This theme was extrapolated from the concepts mapped to Item 17 and responses to Item 9. The mean response for conceptual (M=8.12; SD=3.15) and process (M=.55; SD=.50, t(66)=-20.71,

p<.000). On average, program directors expected more competencies representing conceptual knowledge as compared to process knowledge constructs.

Programs Not Offering Genetic Genomic Content. Thematic analysis used for the program directors whose programs offer genetic content was used for programs that do not provided genetic and genomic content. Although there are programs that do not offer genetic and genomic content, their program directors reported they still expect genetic and genomic competencies in their graduated student (see Table 27). Results of the thematic analysis are that program directors from programs without genetic and genomic content reported; a genetics approach to content is used as compared to a genomic approach, reported basic genetic science principles as compared to genetics and genomics related to discipline-specific content; a more of a theoretical approach to type of knowledge expected is compared to a clinical application of genetic and genomic concepts, and a pedagogic approach to awareness knowledge as compared to process knowledge.

For example on average, program directors that report offering genetic and genomic content in their program, expected genetic based competencies (M=6.00; SD=2.43) from graduated students as compared to those programs who did not offer content (M=4.05; SD=2.13), with a difference in means being 1.95. This trend was noted across all dimensions for all competencies. There was less of a mean difference (.85) in levels of competency (clinical application) across the programs that do and do not offer genetic and genomic content. The largest mean difference between programs that offer genetic content and those that do not was noted in the pedagogic approach (conceptual knowledge). There was a 2.37 mean difference between those programs that offer genetic and genomic content and those that do not.

Table 27. ***Thematic Analysis of Programs With and Without Genetic and Genomic Content***

Rubric Dimensions and Whether Program Offers Genetic and Genomic Content		Frequency of Response N=67	Mean	SD	Mean Difference
Genetic pre-genomic	Yes	47	6.00	2.43	1.95
	No	19	4.05	2.31	
Genomic post-genomic	Yes	47	2.64	1.21	.85
	No	19	1.79	1.82	
Genetic constructs	Yes	47	6.13	1.99	1.99
	No	19	4.16	2.17	
Discipline specific constructs	Yes	47	3.34	1.45	1.23
	No	27	2.11	1.15	
Level of literacy expected	Yes	47	7.36	2.12	2.10
	No	18	5.26	2.58	
Level of competency expected	Yes	42	2.13	1.23	.87
	No	15	1.26	.991	
Conceptual knowledge	Yes	47	8.87	2.92	2.37
	No	18	6.50	2.88	

Summary Query 3. Thematic analysis indicated that programs offering genetic and genomic content tend to take a genetics approach as compared to genomics. The content is likely to be based on principles of basic genetic science as compared to discipline specific. This focus on genetic science logically leads to the result that programs are more likely to focus on theoretical foundations as compared to clinical applications. This is consistent with the result that programs are more likely to measure knowledge by awareness as compared to process knowledge.

Programs that reported they don't offer a genetic and genomic content expected graduated students to exhibit genetic and genomic competencies. However, they expected lower number of competencies than programs that offer genetic and genomic content.

Professional Speech-Language Pathologists

The second objective of this research study was to survey the perceptions of the professional speech-language pathologist in regards to current attitudes, education, knowledge and clinical practices as they relate to genetics and genomics. Results of the survey called, *Genetics in the Post-Genomic Era: A Practitioner's Perspective* are presented in the following order: (1) characterize professional development education in regards to genetic and genomics; (2) characterize the perceptions and attitudes of genetics and genomics; (3) determine the perceived knowledge regarding genetics and genomics as it relates to speech-language pathology; (4) identify the clinical services within scope of practice as it relates to genetics and genomics; and (5) characterize the confidence levels of speech language pathologists as it relates to services performed in clinical practice. Each query result will conclude with a summary.

Professional and Educational Characteristics

Question 24. What is your current certification status?

Of the 215 speech-language pathologists who completed the survey, 97% (208) reported to be ASHA certified speech-language pathologists, while two respondents reported possessing dual certification (certified in both audiology and speech-language pathology). See Table 28 for characteristics of responding speech-language pathologists.

Question 19. Are you male or female?

Ninety-one percent (195) of the respondents were female. The high ratio of females to males is consistent with the gender demographics of the field of speech-language pathology (ASHA, 2010).

Question 17. How long have you been practicing as a speech-language pathologist?

Fifty-three percent of respondents (116) reported being certified as a speech-language pathologist 15 years or less. Thirty-seven percent (59) reported practicing as a

speech-language pathologist for 25 years or more, while 21% (46) reported practicing for 5 years or less.

Table 28. ***Characteristics of Responding Speech-Language Pathologists***

Characteristics of Professional Speech-Language Pathologists	National Demographics*	Percent	Frequency of Response N=215	Percent
Certified members of ASHA	150,241			
Females	144,000	96%	194	91%
Males	6,100	4%	20	9%
ASHA certified speech-language pathologists	130,997	87%	209	97%
Dual certified (audiologist and speech-language pathologist)	1,172	<1%	2	1%

*HES Graduate Guide Survey, 2009-2010 Academic Year

Education in Speech Language Pathology

Question 21. What is your most advanced degree obtained? Sixty-five percent (173/214) of the respondents indicated that their most advanced degree obtained was a *Masters degree*, while and 15% (39) of the respondents indicated a *Doctoral degree* in speech-language pathology was the most advanced degree obtained. None of the respondents indicated they possessed a degree in biology, genetics, or any other genetic related degree.

Question 20. In what year did you complete your most advanced degree? The data was further analyzed to determine the distribution of when the most advanced degree was obtained; during the Pre-Genomic Era or during the Post-Genomic Era. Sixty-three percent (215) of the respondents obtained their most advanced degree during the Pre-Genomic era (before the year 2003). Thirty-seven percent (79) obtained their degree in the Post-Genomic era (after the year 2003). (See Table 29.)

Table 29. Selected Characteristics of Responding Speech-Language Pathologists

Years of experience of responding speech-language pathologists	Frequency of Response N=215	Percent
0-5 years	46	21%
6-10 years	32	14%
11-15 years	38	17%
16-20 years	24	11%
21-25 years	18	8%
26-30 years	24	11%
31+ years	33	16%
Level of education completed	n=212	
Master	173	81%
Doctorate	39	18%
Year completed most advanced degree	N=215	
< 2002	136	63%
2003 to 2011	79	37%

Question 22. Are you currently completing a degree?

Question 24. What degree are you currently completing, and in what area?

Twenty-four speech-language pathology respondents were currently engaged in obtaining an additional degree. Twenty-two of these speech-language pathologists were obtaining a Doctorate of Philosophy degree. The areas of doctoral research reported by the doctoral candidate are contained in Table 30. One respondent indicated conducting research in the area of genetics and or genomics (adult neurogenics). Two respondents indicated they were obtaining a second Bachelors degree in another unspecified field.

Table 30. ***Areas of Doctoral Research***

Area of reported doctoral research	Frequency of response N=215	%	Area of reported research	Frequency of response N=215	Percentage
Speech language pathology	7	35%	Linguistics	1	<1%
Voice disorders	3	15%	Early intervention	1	<1%
Aphasia	2	13%	Human development	1	<1%
Neurogenetics-adult	1	<1%	Interdisciplinary health sciences	1	<1%
Child language	1	<%	epidemiology	1	<1%

Query 1 Genomic Education

Continuing Education in Genetics and Genomics.

Question 3. Have you had any formal education in any topic of genetics or genomics (i.e., developmental embryology, molecular biology, genetics, genomics, etc.). If you answered "Yes", fill out what year you completed such course(s).

The speech-language pathologists were asked to indicate any history of formal genetic and/or genomic education, including certificate degrees, and continuing educational events. Multiple responses were accepted. None of the respondents reported obtaining a degree in genetics or any of its related fields. The educational level and year of continuing education events is provided in Table 31.

Eighty percent (212/265) of the respondents indicated they had taken a course with genetic and genomic content. When asked, at what level of education was the course taken, the following responses were obtained. An educational course(s) with genetic and/or genomic content (either a specific course or integrated content) was taken at the undergraduate level by 51% of the respondents. Forty-four percent of the respondents indicated they had some form of genetic and/or genomic content (described as basic science) in high school, 37% of the respondents had taken course(s) with genetic and or

genomic content at the graduate level, 9 respondents at the post graduate level, thirteen respondents, at the doctoral level, and four respondents at the post-doctoral level.

Table 31. ***Levels of Genetic and Genomic Education***

Level of education with genetic and genomic content	n=	Frequency of Response	Percent
High school	163	71	44%
Undergraduate	173	88	51%
Graduate	165	61	37%
Post graduate	135	12	9%
Doctoral	126	13	10%
Post doctoral	119	4	3%
Educational courses			
Integrated cleft palate course	163	88	54%
Continuing education courses	157	64	41%
Stand-alone genetics and speech-language pathology course	127	13	10%

Fifty-four percent (88/163) of the respondents indicated they received genetic and genomic education that was integrated in a cleft palate/craniofacial course. Forty-one percent (64) of the speech-language pathologists reported attending continuing education events (conferences, seminars, workshops, etc.). Ten percent (13) of speech-language pathologists responding to the survey indicated they had taken a stand-alone genetics and speech-language pathology course. (See Table 32.)

Table 32. ***The Most Recent Year an Education Event was Attended***

Year attended education event with genetics and genomics content N=166	Frequency of response	Percent	Year attended education event with genetics and genomics content	Frequency of response	Percent
2011	18	11%	2004	8	3%
2010	19	11%	2003	4	2%
2009	13	8%	2002	5	2%
2008	15	9%	2001	5	2%
2007	4	2%	2000	10	4%
2006	14	8%	<1999	47	18%
2005	4	2%			

ASHA Sponsored Genetic Genomic Continuing Education Events.

Question 4. Have you attended any ASHA sponsored continuing education events where the content was genetically and/or genomic related? Professional speech-language pathologist were asked if they attended American Speech- Language Hearing Association (ASHA) sponsored continuing education (CEU) events where genetics and genomics were included in the content. Of the 266 responses, 24% (64) speech- language pathologists reported attending an ASHA CEU event in which genetic and or genomic content were offered.

Question 5. In what year did you attend an ASHA CEU event in which genetic and genomics was included in the content of the event? In response to what year were the genetic and genomic education events were attended, responding speech-language pathologists reported that the majority of events were attended since the year 2003. Ninety-five respondents (%) attended an educational event with genetic and genomic content since the year 2003. Seventy-two respondents (%) attended an educational event with genetic and genomic content prior to the year 2003.

Query 1 Summary. The majority of the survey respondents were ASHA certified, female, speech-language pathologists. Fifty-nine percent reported being in practice for 15

years or less, with 21% being in practice of less than 5 years. Eighty-nine percent of the respondents most advanced degree obtained was the Masters degree. None of the respondents reported to have a degree in genetics and or any other related field. Twenty-one of the responding speech-language pathologists were engaged in obtaining a Doctorate of Philosophy. One respondent indicated the obtained degree was in the area related to genetics and genomics.

Eighty percent of the responding speech-language pathologists report attending continuing education events with genetic and genomic content, yet only twenty-four percent of speech language pathologists attended ASHA sponsored events where genetics and genomics were included in the content. Fifty-four percent of the respondents indicated that the educational event with genetic and genomic content was included occurred while taking a craniofacial disorders course. Of those respondents who had enrolled in an educational event with genetic and genomic content, 60% of the respondents had taken the educational event since the completion of the Human Genome Project in 2003 (The Post-Genomic Era).

Query 2. Perceptions of Genetics and Genomics

Awareness of Genetic and Genomic Advancements.

Question 1. Are you aware of any advances in genetic/genomics within the last 5-10 years that directly relate to speech-language pathology? Speech-language pathologists were asked about their awareness of recent advancements (defined as being within the last 10 years) in genetics and genomics that directly relate to speech- language pathology. Seventy percent of the responding speech-language pathologist reported they were aware of recent advancements.

Impact of Genetics and Genomics.

Question 2. In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology? When asked their opinion as to

what impact the current field of genetics and genomics had on speech-language pathology, 37% (99/263) indicated that genetics and genomics had a "significant impact" on speech-language pathology, followed by 35% indicating a "moderate impact" (95), a "minimal impact" by 14%, and one responder indicated the current field of genetics and genomics had "no impact" on speech-language pathology. (See Table 33.)

Table 33. ***Perceived Level of Impact of Genetics and Genomics Reported by Responding Speech-Language Pathologists***

Level of impact of genetics and genomics	Frequency of response N=266	Percentage
Significant impact	99	37%
Moderate impact	95	36%
Minimal impact	33	12%
No impact	1	<1%

Preparedness.

Question 6. In your opinion, how well did your speech-language pathology degree program prepare you to understand the current field of genetics and genomics? Speech-language pathologists were asked their opinion as to how well their speech-language pathology degree training program prepared them to understand the current field of genetics and genomics. Sixty-three percent (166/263) indicated they did "not feel prepared." Thirty-four percent (89) felt "somewhat prepared," five responding speech-language pathologists felt "prepared", and three of the respondents indicated they felt "very prepared" in understanding the current field of genetics and genomics.

Sixty-three percent (166/263) of responding speech-language pathologists reported they felt their degree training program had not prepared them for understanding genetics and genomics, while thirty-six percent (23/65) of responding program directors indicated that graduated students are prepared appropriately for knowledge and skills in genetics and genomics.

Perceptions of Genetics and Genomics and Work Practices.

Question 8. Of the following, which area(s) of communication sciences do you believe genetics and genomics plays a role? Professional speech-language pathologists were asked as to which processes and areas of disorders of communication sciences did genetics and genomics plays a role. Ninety-nine percent of the responding speech-language pathologist reported that genetics and genomics play a role in the areas of autism, hearing, learning, and pediatric articulation. All speech language pathologists who responded to this question, overwhelmingly indicated genetics and genomics plays a role in all of the processes listed. (See Table 34.)

Table 34. ***Speech and Language Processes Reported Where Genetics and Genomics Play a Role***

Speech and language processes	Frequency of responses N=266	Percent	Speech and language processes	Frequency of responses N=266	Percent
Autism	220 (222)	99%	Fluency	208 (218)	95%
Hearing	220 (220)	99%	Pediatric articulation	206 (218)	94%
Learning	219 (221)	99%	Resonance	204 (216)	94%
Pediatric language	217 (220)	99%	Fluency	208 (218)	94%
Attention	216 (220)	98%	Voice	196 (213)	92%
Cognition	218 (222)	98%	Feeding	194 (214)	91%
Reading	206 (211)	98%	Adult language	194 (215)	90%
Physical assessment	206 (218)	96%	Swallowing	187 (213)	87%
Social pragmatics	208 (215)	96%	Adult articulation	184 (215)	85%

Query 2 Summary. Seventy percent of speech-language pathologists indicated they were aware of recent genetic and genomic advancements in speech-language pathology. Approximately 35% of the respondents felt these advancements to have at least a *"moderate impact"* on the field of speech- language pathologists. Sixty-three percent of speech-language pathologists did not feel their speech-language pathology

degree program prepared them to understand current needs of genetics and genomics, whereas less than 1% felt *"prepared"* or *"very prepared."* Responding speech-language pathologists indicated that genetics and genomics play a role in all of the processes of communication and its disorders.

Query 3. Knowledge of Genetics and Genomics

Question 16. Do you have the knowledge to answer the following questions? Survey participants were asked their perceptions of their ability to answer specific questions related to genetics and genomics in the field of speech-language pathology. The respondents were asked to indicate their level of knowledge of 20-items based on a 4-point Likert-scale (1-no knowledge, 2-some knowledge, 3-significant knowledge, and 4-expert knowledge). The items reflected both principles of basic genetic science and principles of genomics as they both specifically relate to communication sciences and its disorders. Results indicated that speech-language pathologists overwhelmingly felt they had "no knowledge" in at least 85% of the constructs. Table 35 depicts the ranked frequency and percent of responses.

Result Summary Query 3. Professional speech-language pathologists reported their perceived knowledge of genetic and genomic basic principles and discipline specific constructs in speech language pathology to be poor with the majority indicating they have "no knowledge" in the constructs. Sixty percent of the respondents reported a degree of knowledge in the presented constructs. Results revealed a tendency to "know" more genetic related knowledge constructs than genomic related constructs.

Table 35. ***Perceived Knowledge of Genetic and Genomic Constructs***

	Percent and frequency of response of speech-language pathologists N=218				
Questions	No knowledge	Some knowledge	Significant knowledge	Expert knowledge	N
1.What is GINA?	94% (202)	5% (10)	1% (2)	<1% (1)	215
2. What is the significance of GJB2 gene?	92% (198)	7% (12)	2% (5)	0%	215
3. What is the significance of CNTNAP2?	93% (196)	3% (7)	4% (9)	0%	212
4. What is a candidate gene?	83% (177)	12% (26)	4% (9)	1% (4)	216
5. What does FISH stand for?	74% (160)	18% (39)	7% (15)	1% (3)	217
6. What is the significance of FOXP2?	74% (157)	18% (39)	8% (18)	0%	214
7. What are the legal ramifications of a genetic disorder in the educational system?	74% (157)	21% (45)	4% (8)	1% (3)	213
8. Do you know how to take a family pedigree history?	70% (151)	26% (57)	3% (6)	1% (2)	216
9. Are speech language disorders polygenic or monogenic?	72% (152)	18% (39)	9% (20)	<1% (1)	212
10. What are modes of non-traditional Inheritance?	71% (151)	26% (57)	3% (7)	0%	214
11. What is epigenetics?	70% (149)	23% (50)	6% (12)	1% (3)	214
12. What is karotype analysis?	68% (144)	27% (58)	4% (9)	1% (2)	213
13. What is the inheritable rate of speech language disorders?	68% (146)	27% (58)	4% (9)	<1% (1)	214
14. Name 10/40 metabolic disorders that are typically screened at birth?	65% (140)	31% (67)	2% (5)	1% (4)	215
15. What are linkage and association studies?	65% (140)	28% (60)	7% (14)	<1% (1)	215
16. What is the genetic relationship to GERD feeding disorders?	62% (133)	31% (65)	6% (13)	1% (2)	213
17. What is the genetic relationship to apraxia?	61% (132)	35% (75)	42% (9)	0%	216
18. What is the susceptibility rate of stuttering disorders?	42% (95)	38% (82)	15% (32)	3% (6)	215
19. What is the difference between a chromosome anomaly and a single gene defect?	41% (88)	46% (99)	12% (25)	2% (4)	216
20. When does one refer to a geneticist?	39% (84)	46% (98)	13% (27)	2% (5)	214

Query 4. Work Practices of Speech-Language Pathologists

Clinical Setting.

Question 25. Are you currently engaged in clinical practice? Eighty-three percent (181/217) of the respondents indicated they were engaged in clinical practice at the time of answering the survey.

Question 27. What is your primary work setting? Twenty-five percent (53) of the responding speech-language pathologists reported to work in a hospital setting, 21% reported working in an elementary school setting, and 19% reported working in an academic setting. Table 36 provides the ranked frequency and percent of all reported work settings. As reported in 2009, of the 145,000 members, 57% of certified speech-language pathologists were employed in an educational setting, 38% were employed in the healthcare setting, and 15% of certified speech-language pathologists were employed in non-residential healthcare settings (ASHA, 2012).

Table 36. ***Primary Work Setting of Responding Speech-Language Pathologists***

Type of work setting	Frequency of response N=214	Percent	Type of work setting	Frequency of response N=214	Percent
Hospital	53	29%	Preschool	16	7%
Elementary school	44	21%	Early intervention	12	6%
Academic setting	39	19%	Middle and high school	8	4%
Private practice	33	16%	Nursing home	3	1%
Rehabilitative centers	18	9%	Corporate setting	3	1%

Area of Specialty.

Question 26. What is/are your specialty area(s) of practice? Respondents were asked to indicate their area(s) of practice and/or specialty. Multiple responses were accepted. Forty-seven percent (100/214) of responding speech-language pathologists

indicated pediatric language disorders as their area of practice. This was followed with the specialty area of general speech language pathology being reported by 38% of the respondents, while 34% indicated dysphagia and autism as areas of specialty practice (see Table 37).

Table 37. ***Areas of Practice as Reported by Responding Speech-Language Pathologists***

Area of practice	Frequency of response N=213	Percent	Area of practice	Frequency of response N=213	Percent
Language disorders-pediatric	100	46%	Early childhood feeding	33	15%
General speech-language pathology	82	38%	Head and neck cancer	33	15%
Dysphagia	73	34%	Intellectual disability	30	14%
Autism	69	34%	Bilingualism	29	14%
Phonological disorders	70	33%	Auditory processing	28	13%
Pragmatic language	63	29%	Resonance disorders	28	13%
Apraxia of speech	57	27%	Craniofacial speech disorders	27	13%
Motor speech disorder	56	26%	Augmentative Ccommunication	26	12%
Cognitive communication	54	25%	Infant feeding	25	12%
Voice	53	25%	General pediatrics and adults	23	11%
Aphasia	50	24%	Prelinguistic vocal development	18	8%
Language disorders-adult	47	22%	Neuroscience	16	7%
Specific language impairment	46	22%	Oral myofunctional	15	7%
Fluency disorders	46	22%	Hearing impairment	14	7%
Learning disabilities	43	20%	Accent reduction	14	7%
General adult	42	20%	Aural rehabilitation	9	4%
Literacy	35	16%	Public speaking	8	4%

Relationship to Craniofacial Teams and Centers.

Question 31. Are you currently or have been in the past been a member of a craniofacial and or cleft palate team/center? Working in a setting where genetics and genomics are an integral component of clinical service, where recent genetic and genomic research findings are readily accessible, and there are opportunities to collaborate with geneticists and genetic counselors, provides the speech-language pathologist a unique opportunity of being more knowledgeable and familiar with genetic and genomics. Twenty-two percent (48/214) of the respondents were reported to be a member of a craniofacial team and or center.

Question 33. Was a geneticist or genetic counselor present at the team meetings at least 80% of the time? Twenty-two responding speech-language pathologists reported that a geneticist was not present at team meetings. Sixteen respondents reported a geneticist was present at the meetings, while six responding speech-language pathologists did not know if a geneticist was in attendance at the meetings.

Due to the nature and access to genetic and genomic information being a member of a craniofacial team may afford, question was raised whether there was a relationship between perceived level of knowledge of genetics and genomics and being a craniofacial team member. Speech-language pathologists who were craniofacial team members indicated more perceived *knowledge* (*M*=47.42; *SD*=28.53) in genetic and genomic constructs than those who were not a member of a craniofacial team (*M*=24.61; *SD*=23.75, *t*(197) =5.37, *p*=.001). The distribution was highly skewed. The Mann-Whitney U test was conducted and revealed similar results as the independent samples t-test.

Speech-language pathologists who reported to be craniofacial team members also reported more perceived knowledge in *genetic related knowledge* constructs (*M*=48.86; *SD*=28.14) as compared to non team member respondents (*M*=23.68; *SD*=24.07, *t*(107)=5.89, p=.001). This trend was also noted for those responding speech-language pathologists (*M*=40.23; *SD*=28.14) who reported more perceived knowledge in *genomic*

related knowledge constructs (*M*=23.16; *SD*=23.85, *t*(196)=3.99, *p*=.001). (See Table 38.) In all three areas, speech-language pathologists who reported being a member of a craniofacial team and or center perceived more knowledge of genetic and genomic constructs than those speech-language pathologists who reported that they were not a craniofacial team member.

Teaching Practices.

Question 28. Are you teaching in the field of speech-language pathology? Working within an academic setting where one may have access recent research findings, as well have opportunities to collaborate with fellow faculty members and students, may provide the speech-language pathologist a unique opportunity for increased exposure to genetics and genomics. Twenty-nine percent (62/213) of the respondents indicated they are teaching in the field of speech-language pathology.

Question 29. What academic level and what course(s) do you teach? Eighty-seven percent (44/54) are teaching at the graduate level, while 26 speech-language pathologists reported teaching at the undergraduate level. Six of responding speech-language pathologists reported teaching at the post-graduate level. A list of the reported courses is located in Appendix S.

Analysis was performed on whether teaching in an academic setting had an effect on perceived level of knowledge and level of confidence. (See Table 39.) The distribution was highly skewed. The Mann-Whitney U test conducted and revealed similar results as the independent sample t–test. Speech-language pathologists who reported teaching in an academic setting reported more knowledge (M=36.07; SD=25.54) than those who do not teach (M=26.49; SD=26.15, t(197) =2.30, p=.022).

Analysis revealed differences between the response rate means of teaching speech language pathologists, and those respondents who reported to not teach, on the question probing perceived genetic related knowledge (teaching SLPs: M=33.52; SD=25.71;

Table 38 ***Association Between Perceived Knowledge and Craniofacial Team Member***

	Craniofacial team member	Frequency of response	Mean	*SD*	*t*	*df*	*Sig.*	Size of discrepancy*
Percent speech-language pathologists with any knowledge of constructs	Yes	44	47.42	28.53	5.37	197	.000	22.81
	No	155	24.61	23.75				
Percent speech-language pathologists with any knowledge of genetic constructs	Yes	44	48.86	28.14	5.89	107	.000	15.18
	No	155	23.68	24.07				
Percent speech-language pathologists with any knowledge of genomic constructs	Yes	43	40.23	28.24	3.99	196	.000	37.07
	No	155	23.16	23.85				

Table 39. ***Association Between Perceived Knowledge and Teaching***

	Teaching in the field of speech-language pathology	Frequency of response	Mean	*SD*	*t*	*df*	*Sig.*	Size of discrepancy
Percent speech-language pathologists of constructs with any knowledge	Yes	54	36.07	25.54	2.30	197	.022	9.58
	No	145	26.49	26.15				
Percent of speech-language pathologists knowledge of genetic constructs	Yes	54	33.52	25.71	1.52	107	1.29	6.49
	No	145	27.03	27.06				
Percent of speech-language pathologist knowledge of genomic constructs	Yes	53	34.15	25.90	2.64	196	.009	10.63
	No	145	23.52	24.80				

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non-teaching SLPs: M=27.03; SD=27.06, t(107)=1.52, p=1.29). However, this finding there was no difference when an independent t-test was performed. Results reported a relationship between those who teach and their perceived genomic related knowledge constructs (M=34.15; SD=25.90) and speech language pathologist who do not teach (M=23.52; SD=24.80, t (196)=1.06, p=.001).

Question 30. Are you currently conducting research that involves genetics or genomics? Ninety-eight percent of the responding speech-language pathologists reported they are not currently conducting research in genetics or genomics. Five respondents reported they are engaged in research. The following areas of research were reported: "22q11.2 deletion syndrome," "Motor Speech Disorders," "Stuttering and fluency disorders," "Genes related to cognitive development, 5HTT, etc.," and "Transgenic models of neurodegenerative diseases (mostly Parkinson disease)."

Clinical Activities Routinely Performed.

Question 12. Of the following activities involved in the assessment and management of speech-language development and disorders, indicate which activity(ies) you have or are currently performing and the level of confidence you feel you have for each activity. Speech-language pathologists were asked what genetic and or genomic clinical services they perform within their work setting. Approximately half of the respondents indicated they perform at least one of the genetic and genomic related clinical activities. Fifty-three percent (111/208) of speech-language pathologists reported *discussing the genetic nature of a speech-language disorder* in their work practices, followed by 38% (77) of the respondents reporting *making referrals based on suspect assessment*, and 44% (80) of the responding speech-language pathologists reported they engage in *eliciting a family history* as a component in their work practices. Table 40 provides the complete list of the genetic and genomic clinical services performed by responding speech- language pathologists.

Table 40 ***Genetic and Genomic Related Activities Performed in Clinical Practice***

Clinical genetic and genomic practices performed by speech-language pathologists	Number and percent who perform activity	
	Frequency of Response N=215	Percent
1. Discuss the genetic nature of a speech language delay or disorder	111/208	53%
2. Make a referral for genetic assessment and counseling	77/201	38%
3. Elicit a genetic family history	80/206	44%
4. Counsel on characteristics of genetic speech language disorders	77/201	38%
5. Correct misconceptions about genetic disorders	77/199	38%
6. Assess the genetic contribution to a speech language disorder	73/207	35%
7. Counsel on impact of speech language disorder on family	59/200	30%
8. Discuss legal, ethical and social issues	57/200	29%
9. Discuss whether genetic testing should occur	51/202	25%
10. Possible prognosis (susceptibility and recurrence risks)	36/197	18%
11. Discuss modes of inheritance	34/202	17%
12. Perform a pedigree assessment in regards to genetics and speech language pathology	18/200	9%
13. Provide a genetic speech language diagnosis	15/199	8%
14. Counsel on types of genetic testing	11/198	6%

The clinical skills achieved by a professional speech-language pathologist are an accumulation of opportunity, experience and knowledge. The education provided in a training degree program provides a foundation of the theoretical framework and competencies needed in clinical practice. Determining the association of program directors expected competencies of graduated students and the type of clinically related competences typically engaged by the professional speech-language pathologist would characterize the type of genetic and genomic competencies typically used in practice. It provides information on what type of competencies should be integrated into curricula in degree training programs.

The genetic and genomic clinical services reported by responding speech-language pathologists were compared to the responding program director's expected competencies of their graduated students. Responding speech-language pathologists responded "Yes" if they performed the clinical service and "No" if they did not perform the activity. Responding program directors indicated "Yes" if expected the competency (or activity) from their graduated student or "No" if they did not expected the competency (or activity) from their graduated student. (See Table 41.)

Table 41. ***Comparison of Genetic and Genomic Activities Performed and Competencies Expected of Graduated Students***

Activity performed in clinical practice (n=215) and expected by graduating students (n= 68).		Yes		No	
Discuss the genetic nature of a speech language delay or disorders	SLP	111	53%	97	47%
	PD	16	24%	51	76%
Make a referral for genetic assessment and counseling	SLP	93	45%	114	55%
	PD	50	75%	17	25%
Elicit a genetic family history	SLP	90	41%	116	56%
	PD	21	31%	46	69%
Counsel on characteristics of genetic speech language disorders	SLP	77	38%	124	62%
	PD	58	87%	9	13%
Correct misconceptions about genetic disorders	SLP	77	39%	122	61%
	PD	34	51%	33	49%
Assess the genetic contribution to a speech language disorder	SLP	73	35%	134	41%
	PD	26	39%	41	61%
Counsel on impact of speech language disorder on family	SLP	59	29%	141	71%
	PD	33	49%	34	51%
Discuss legal, ethical and social issues	SLP	57	28%	143	72%
	PD	5	8%	62	93%
Possible prognosis (susceptibility and recurrence risks)	SLP	36	18%	161	82%
	PD	31	46%	36	54%
Discuss modes of inheritance	SLP	34	17%	168	83%
	PD	35	52%	32	48%
Provide a genetic speech language diagnosis	SLP	15	8%	184	93%
	PD	36	54%	31	46%
Counsel on types of genetic testing	SLP	11	6%	187	94%
	PD	4	6%	63	94%

Of the 12 items offered, responding program directors expected more of the activities as demonstrated competencies from their graduated students in comparison to the activities reported to be performed by responding speech-language pathologists. Table 42 presents the percent difference between the program directors and speech-language pathologists. In nine of the constructs offered, program directors reported to expect more than activities than the speech-language pathologists. Speech-language pathologists reported they perform the 10 items versus two items represented areas of clinical practice.

Table 42. ***The Percent Difference in Genetic and Genomic Activities Expected From Graduated Students and Activities Performed by Speech-Language Pathologists***

Activities Expected More from Program Directors (n=) as Competencies from Graduated Students	Percent Difference in the Reported Activities Expected	Activities Performed More by Speech-Language (n=) than Expected as Student Competencies from the Program Directors	Percent Difference in the Reported Activities Performed
Counsel on characteristics of genetic speech-language disorders	49%	Discuss the genetic nature of a speech language delay or disorders	20%
Provide a genetic speech language diagnosis	47%	Discuss legal, ethical and social issues	20%
Discuss modes of inheritance	35%	Elicit a genetic family history	10%
Make a referral for genetic assessment and counseling	30%	Counsel on types of genetic testing	0
Possible prognosis (susceptibility and recurrence risks)	28%		
Counsel on impact of speech language disorder on family	20%		
Correct misconceptions about genetic disorders	12%		
Assess the genetic contribution to a speech language disorder	4%		
Counsel on types of genetic testing	0		

The results report that professional speech-language pathologists are not performing the type and amount of genetic and genomic related clinical activities that are expected as competencies in graduated students.

Interest in Genetics and Genomics Expressed by Patients/Clients/Family.

Question 9. Have you received questions from parents/patients/clients regarding genetic/genomic principles of basic science such as ways in which conditions or traits are inherited, risks for recurrence of a condition or trait, etc?

Question 10. Have you received questions from parents/patients/clients regarding genetic/genomic and speech-language pathology?

Respondents were asked if they had received questions from their patients, clients, and or family members about basic genetic science, as well as questions regarding genetics and speech- language disorders and or delays. Speech-language pathologists indicated that 59% received questions regarding basic genetic science and 53% of the questions pertained to genetics of speech-language pathology disorders and or delays.

Referral Practices.

Question 15. To which professional would you FIRST refer if you suspected a genetic/genomic relationship occurring in a patient/client? Table 43 provides the referral practices of the responding speech-language pathologist. Fifty-three percent (112/212) of responding speech-language pathologists would make a referral to a pediatrician or developmental pediatrician. This was followed by 17% (35) of responding speech-language pathologists referral to a geneticist, thirty-five referring to a genetic counselor, while twenty-seven responding speech-language pathologists indicated referring to a neurologist. Fifteen of the respondents indicated they would not make a referral upon recognizing a possible genetic relationship to a disorder or condition.

Descriptive analysis of the textual comments (See Appendix R) indicated that 5 respondents indicated that although they would like to make a referral if required, they are unable to do so because of school district policy. Ten respondents indicated that

referral practices were dependent on the type of insurance coverage held, and 15 respondents (indicated they would make a referral to a primary care physician.

Table 43. ***Referral Practices of Speech-Language Pathologists***

Type of Specialist Typically Used to Refer an Individual Suspected of a Genetic Related Speech Language Condition or Disorder	Frequency of Response N=212	Percentage
Pediatrician or developmental pediatrician	131	62%
Geneticist	35	17%
Genetic counselor	35	17%
Neurologist	27	13%
Primary care physician	15	7%
Would not make a referral	13	6%

Query 4 Summary. Responding speech-language pathologists indicated that they primarily work in a hospital or elementary school setting, with the majority practicing in the area of pediatric language disorders and general pediatric speech-language pathology. Twenty-two percent of responding speech-language pathologists indicated they were or had been a member of a craniofacial team, and twenty-eight percent indicated that they also engage in teaching within the field of speech- language pathology. Approximately half of the speech-language pathologists reported to engaged in genetic related clinical services as compared to genomic related clinical services (primarily in the provision of management and counseling services). A little over fifty percent received questions about the science of genetics and or specific questions related to genetics and speech-language disorders from individual patients and or their family. Sixty-two percent would refer to a pediatrician if a genetic issue were suspected. Significant differences were found in the types of clinical services typically engaged in by professional speech-language pathologists and the type of expected competency expected of graduated students by program directors.

Query 5. Confidence Levels in Genetics and Genomics

Respondents were asked to rate their level of self-confidence (on a 4-point Likert scale (1-"*not confident,"* 2-"*somewhat confident,"* 3-"*confident,"* 4-"*very confident"*) in constructs related to typical clinical services of a speech-language pathologist. These activities included; basic foundational understanding of genetics and genomics, assessment, diagnosis, management, and counseling (Items 7, 8, 9, 11, 12, 13, 14, and 15).

Confidence in Understanding Genetics and Genomics.

Question 7. Do you feel confident in your understanding of the current field of genetics and genomics as it applies to the field of speech-language pathology? Fifty-six percent (147/265) of the responding speech language pathologists reported their perceived confidence levels in understanding of current genetics and genomics as it relates to speech language pathology as "*not confident."* While 37% (97) reported they were "*somewhat confident,"* fifteen responding speech-language pathologists reported they were *"confident,"* and six respondents indicated they were *"very confident"* in understanding genetics and genomics as it relates to speech language pathology.

A Pearson Chi-square test of independence was performed to examine the relationship between responding speech-language pathologist's perceived knowledge of genetic and genomic constructs and perceived levels of confidence. The relationship between these variables was significant, χ^2 (1)=.642, p=.001. Speech-language pathologists were more likely to perceive more confidence when they perceived more knowledge of genetic and genomic constructs. This trend was similar whether the knowledge constructs were related to *genetics* (χ^2(1)=.625, p=.001) or to *genomics* (χ^2(1)=.583, p=.001).

Confidence Levels of Various Clinical Practices.

Question 13. How confident are you in providing counseling/information on genetic/genomic speech-language pathology issues? Sixty-two percent (110)

respondents reported they were not confident in providing information and or counseling on speech-language pathology issues. Table 44 provides the levels of confidence reported by the remaining respondents who reported any degree of confidence.

Table 44. ***Level of Confidence Reported by Speech-Language Pathologists***

Level of Confidence in Providing Information and or Counseling	Frequency of response N=110	Percent
Not confident	140	62%
Somewhat confident	71	32%
Confident	13	6%
Very confident	1	<1%

Question 11. Please rate your level of confidence in knowing you were providing the appropriate genetic/genomic information/answers to questions posed by parents/patients/clients regarding genetics and genomics. Sixty percent (136/228) of responding speech-language pathologists reported they received questions about basic genetic science from their patients/clients and or family members. Forty-seven percent of responding speech-language pathologists indicated they were *"somewhat confident"* in knowing they were providing appropriate information in their answers. Thirty-eight percent of responding speech-language pathologists indicated they were *"not confident"* in their responses to the questions posed of the patients/clients and or family members. The levels of *"confident"* and *"very confident"* were reported by twenty-five respondents and five of responding speech-language pathologists, respectively.

Question 12. Of the following activities involved in the assessment and management of speech-language development and disorders, indicate which activity(ies) you have or are currently performing and the level of confidence you feel you have for each activity. Among the varied clinical services within the scope of practice of a speech-language pathologist, practices of assessment, diagnosis,

management and counseling are considered benchmarks. Speech-language pathologists were asked to indicate if they performed the presented activity and were asked to (via a 4-point Likert scale) rate their self perceived confidence in the assessment, diagnosis, management and counseling of 15 offered genetic and genomic constructs. For example, questions included, *"Discuss modes of inheritance?"* or *"Discuss possible prognosis in regards to susceptibility and recurrence risks?"*

Respondent's responses and levels of confidence mapped to various clinical contexts and survey items, along with ranked response frequencies and percentages, are shown in Table 45.

Table 45. ***Genetic and Genomic Related Activities and Level of Perceived Confidence***

Genetic and Genomic Activities Performed Clinical Practice of the Speech-Language Pathologist	Number and Percent Who Perform Activity n=121		Number and Percent "Confident" in Performing Activity n=121	
	f/n	Percent	f/n	Percent
Discuss the genetic nature of a speech-language delay or disorder	111/208	53%	75/140	54%
Make a referral for genetic assessment and counseling	77/201	38%	43/125	34%
Elicit a genetic family history	80/206	44%	29/121	24%
Counsel on characteristics of genetic speech-language disorders	77/201	38%	48/114	42%
Correct misconceptions about genetic disorders	77/199	38%	53/120	44%
Assess the genetic contribution to a speech-language disorder	73/207	35%	56/115	49%
Counsel on impact of speech-language disorder on family	59/200	30%	39/105	37%
Discuss legal, ethical and social issues	57/200	29%	35/100	35%
Discuss whether genetic testing should occur	51/202	25%	29/94	31%
Possible prognosis (susceptibility and recurrence risks)	36/197	18%	31/90	34%
Discuss modes of inheritance	34/202	17%	31/92	34%
Perform a pedigree assessment in regards to genetics and speech-language pathology	18/200	9%	17/81	21%
Provide a genetic speech-language diagnosis	15/199	8%	13/79	17%
Counsel on types of genetic testing	11/198	6%	13/76	17%

Fifty-three percent (111/121) of the respondents indicated they "*discuss the genetic nature of a speech language delay or disorder with their patient/client and or the parent/family member"* (the highest ranked activity). However, only 18% (24/140) of the responding speech- language pathologists indicated they were "confident" in performing that activity.

Query 5 Summary. Although speech-language pathologists reported to perform clinical activities involving genetics and genomics, less than half of the respondents indicated confidence in performing those activities. Speech-language pathologists reported being more confident in providing referral and counseling procedures as compared to diagnostic and assessment procedures. Levels of confidence in understanding genetics and their perceived knowledge of genetic and genomic constructs as they relate to speech-language pathology are low. Responding speech-language pathologists who are members of a craniofacial team and/or engage in academic teaching perceived their confidence in their knowledge of genetic and genomic constructs to be higher than those responding speech-language pathologists who are not team members or do not teach.

Chapter V

DISCUSSION

Genomics is a young and rapidly evolving field. However, we have not yet determined either the extent of genomic knowledge needed, nor the manner of clinical application of that knowledge (Guttmacher, Porteous & McInerney, 2007). Educating both those in practice and those in training about key concepts of genomics will assist in determining the knowledge needed in order to become genomically literate and competent. Engaging educators and professional speech-language pathologists in the design of effective delivery models for this knowledge will facilitate the translation of genomic science to clinical practice. It is important to teach the key underlying concepts of genomics in communication disorders, and to instill an appreciation of the future clinical importance of genomics. This will motivate students and professional speech-language pathologists to be lifelong learners of genetics and genomics.

One goal in educating the speech-language pathologist in genomics is to provide tools that can be used immediately. The role of the speech-language pathologist is to accurately identify those who require evaluation, determine areas of need, and provide management and treatment when necessary. However, with enhanced dissemination of advances in genomics, more and more non-syndromic individuals will present to the speech-language pathologist with a recognizable genetic and/or genomic component – a

"genomic profile". A genomic profile is a composite of genetic, environmental factors, and their interactions that contribute to the speech and/or language disorder in question. A genomic profile can provide a basis from which a relevantly-trained speech-language pathologist can derive information that will assist in determining causation, establishing a diagnosis, describing the manifestation of the communication disorder, and guiding evidence-based management protocols.

An example of the importance of genomic profiling comes from the area of hearing impairment. For isolated hearing impairment, many believe that if there is no family history, the hearing impairment is not genetic. Therefore, assessment and treatment of a hearing impairment may be narrowly defined, focusing on the management of symptoms currently being presented by the patient/client. However, with current genomic knowledge, one would understand that in fact, 50% of all hearing impairment is due to purely genetic factors, and most occur without a family history. The possibility that hearing impairment is genetic is also evident in the fact that normal hearing, healthy parents of a hearing impaired child have a significantly elevated risk for having a second hearing-impaired child, approximately 1 in 6 (Robin, 2008). Since 2003, advances in genetic testing have enabled further refinement of these risks. In children and adults with northern European descent, for example, there is a 30% likelihood that the child's hearing impairment is due to a connexin 26 gene mutation. If positive, this results in the parents having a 1 in 4 risk of having hearing-impaired child with a future pregnancy. If negative, that risk is only reduced to 1 in 7, reflecting the fact that hearing impairment is genetically heterogeneous, indicating many other candidate genes underlying a child's hearing impairment. This genomic knowledge adds to the potential "genomic profile" of

an individual. Establishing a genomic profile translates to possible changes in the typical diagnostic routines, and changes in management. Robin (2008) reports increasing referrals to geneticists for further evaluation and testing to determine the pathogenesis of the hearing-impairment, thus changing prevalence rates of genetic related hearing impairment.

Similar paths in understanding the pathogenesis of speech and language disorders are currently unfolding. For example, recent research in specific-language impairment has further defined the phenotypic characteristics associated with the disorder. It is defined as persistent language impairment with normal intelligence and in the absence of any explanatory medical conditions. There is an overlap with speech sound disorders, dyslexia, and attention deficit hyperactivity disorder. It is associated with forkhead box P2 (FOXP2), contactin-associated protein like (CNTNAP2) on chromosome 7, calcium-transporting ATPase 2 (ATP2C2), and c-MAF inducing protein (CMIP) on chromosome 16q (Newbury, Simeon, & Monaco 2010). The 8% recurrence risk in preschool children is thought to be high in comparison to the recurrence risk of developmental language disorders. The principal risk factors for specific language impairment were parents who had greater rates of learning problems, especially learning disorders in the mothers and speech and learning disorder in the fathers (Tomblin, Smith & Zhang, 1997). The parents of the children with specific language impairment were also more likely to smoke tobacco during and after the study child's birth than the parents of the control children. Four candidate genes, heritability, shared environment as well as other yet defined factors illustrate the genomic relationship with specific language impairment, genetic and environmental. At present, the pathogenesis of specific-language impairment is not fully

understood (in part, due to the lack of recognition that this type of language disorder have a genomic underlying cause). Yet, research indicates that specific-language impairment manifests with a specific phenotype. However, on initial presentation, the disorder presents as other common language disorders of receptive and expressive deficits. It is only when one looks beyond the "obvious," using a profile from which to anchor a phenotype, does one suspect other causal factors such as genomics.

To provide leading-edge, high quality services, speech-language pathologists need to keep pace with genomics related scientific advances and discoveries and to incorporate these findings into practice. For example, speech-language pathologists will need to recognize when genomic factors play a role in risk and susceptibility of delays and disorders, and respond by making appropriate recommendations. In their role as counselors and educators, speech-language pathologist must be prepared to help patients comprehend the implications of genetic information; answer questions and address concerns related to genetic complex traits issues; and provide appropriate resources and referrals. Many of these roles are logical extensions of current speech-language pathology practices, although some of them are likely to require more extensive education and training in order for speech-language pathologists to achieve the desired level of competence. These roles will need to be assumed with attention to privacy and confidentiality, ethical issues, and with sensitivity to the impact this information may have on patients and their families from psychological, social, economic, and legal perspectives. An understanding of the basic principles and issues related to genetic and genomic content will provide practitioners with a base for interpreting and applying the

outcomes of genomic research, as well as for delivering maximally effective intervention strategies once genomic care is feasible on a wider scale.

Genetic and Genomic Education

There is a need to provide a means to assess learners' reactions and the acquisition of knowledge of both genomic literacy and competency. The level of genomic literacy among students, faculty, program directors and professional speech-language pathologists is unknown, and no validated measure of that knowledge is available. Designed to distinguish students who understand basic concepts from those who do not, concept inventories are increasingly used in science and technology education, although none have been developed for speech-language pathology.

Self-directed, lifelong learning based on personal experience is an acknowledged, important part of the continuous learning required by practicing speech-language pathologists to maintain their competence over a lifetime of practice. Creating opportunities to facilitate the process of self-directed learning for practicing speech-language pathologists is the mandate of continuing education (CEU) events. Developing programs that are practice relevant, readily accessible, and easy to use is a challenge for CEU research and development.

Gaps in Genomic Education within Degree Programs

The results of this study suggest that graduate programs in speech-language pathology are making efforts to provide their students with a foundation in genetic or genomic concepts. This is occurring without any curricular requirements having been set forth by the Council on Academic Programs in Communication Sciences (CAPSD) and

the Council on Academic Accreditation in Audiology and Speech-Language Pathology (CAA), and without any state regulations, formal genomic educational training, or other guidance as to what genetic and genomic content is pertinent at the entry level. However, several gaps were noted:

1. Program directors expect a degree of genetic literacy and competency from students, but programs are not providing the means by which students can achieve entry-level genomic literacy and competency. Hence graduated students feel unprepared for genomically-informed clinical practice in a world of increased genomic understanding.

2. As per program directors' perceptions, the role of speech-language pathology in genetics continues to focus on the assessment and management of single gene disorders as compared to the contemporary approach of providing a genomic approach to speech and language processes and its disorders. Continued dissemination of this perspective widens the gap in understanding contemporary approaches to genetics and genomics.

3. Although attempts are being made to provide genetic and genomic content in program curricula, there appears to be a lack of consensus regarding what to teach and how to teach it.

a. The quantity and quality of genetic and genomic content has not been systemically assessed across degree programs offering content. However it appears to be less than optimal, based on the perceptions of knowledge by responding speech-language pathologists.

4. Perceived and actual genetic and genomic literacy of students entering in a degree program, and levels of knowledge upon graduating from a speech-language

pathology program, remain unknown. Since 1996, National Science Education Standards have recommended that genetic concepts be included in grade and high school curriculum (Center for Science, Mathematics, and Engineering Education, 1996). Providing genetic and genomic content in graduate training programs should take into account a priori levels of genomic knowledge to best provide a genomic – and discipline specific education at the graduate level.

Gaps in Genomic Awareness for the Professional Speech-Language Pathologist

Discrepancies between educational development of the professional speech-language pathologist and the demands of genomics-related work practices highlight areas of need:

1. The translation of genetic and genomic science into clinical application appears to be less than optimal for the needs of the practicing speech-language pathologist. Education development opportunities with regard to genomics in speech-language pathology (although not systematically assessed here) appear to be insufficient to meet the demand.

2. Speech-language pathologists perceive their roles with regard to genetics and genomics as focused on the assessment and management of single gene disorders as compared to the contemporary approach of providing a genomic approach to speech and language processes and its disorders.

3. Gaps exist in perceived and actual genetic and genomic knowledge of both principles of basic genetic science and discipline-specific genomics. To date, there has not been any published information on evidenced-based criteria as to what degree of genomic knowledge is necessary for practicing speech-language pathologists.

4. There exists a gap, though not yet well defined, between the demands of the public (individuals with communication disorders) and the quality of genomic related services speech-language pathologists may be offering. By viewing communication disorders from a genomically-informed perspective, speech-language pathologists may become aware of approaches for assessment and management that are less apparent under a more traditional, single-gene framework.

The Future of Genomic Education

The ongoing discoveries of how genomics offers avenues for clinical applications in speech-language pathology require clinicians to be sufficiently knowledgeable in genomics to understand when it should be applied, how it should be applied and how to communicate effectively the benefits and limitations. This calls for enhanced genomic education. Efforts have been taken to improve genomic literacy of health care professionals such as physicians, occupational therapists, nutritionists, nurses and physician assistants, but similar efforts have yet been directed at speech-language pathologists. Ferro & Green (2011) describe genomic education efforts thus far as the "push approach". Increasingly structured and rigorous educational content demands are confronting clinicians and educators for maintaining professional education. Many educators recognize that aspects of genomics should be integrated across the continuum of training of clinicians; however, the opportunity cost of any curricular choice, together with pervasive misconceptions about genetics and genomics, present steep barriers to effective change.

What is suggested is transforming a push approach to a "pull", in which educators and clinicians demand genomic literacy and competency (Ferro & Green, 2011). This

requires a focus on establishing unambiguous evidence that links the use of genomic information to improved patient-centered outcomes. Without evidence of clinical utility, all educational efforts are likely to be short-lived with limited effects.

Evidence of clinical utility will also facilitate the development of guidelines by professional organizations, which will provide further direction for curricular changes to include genetic and genomic content areas in degree programs, and at all levels of speech language pathology education, from undergraduate and graduate training through to continuing education for professional development.

Genomic Curriculum

The science has to be made accessible, for instance by seeking clear examples to highlight important concepts. There is a need to develop a curriculum to address the varying needs of different sectors of the community (educators, students, professionals, public) and varying levels of genomic literacy and competency. A critical task is the search for creative teaching techniques and materials. One suggestion is to structure a curriculum as a set of modules. The modular approach allows for greater flexibility in the presenting and selecting of topics that are best suited to the needs of the speech-language pathologist and the educator. Educational methods could include web-based modules platforms with the pre-existing speech language pathology curricula. Web-based video lectures/discussions, and interactive activities (hands-on) involving case studies and role-playing are suggested. Case-base and problem-based learning methods are these methods of delivery genomic education that have been advocated by genetics education specialists (Burton, 2003; Reigert-Johnson, Korf, Alford, Broder, Keats et al. 2004; Korf, 2002).

Supporting Genomic Education at the National Level

A long-term phased educational policy, inclusive of students, educators, and their institutions, should be a goal for inclusion of genetic and genomic content training in speech-language pathology. This goal would require collaboration among national organizational departments and committees of statutory and regulatory bodies, universities and colleges, and professional associations. The engagement of regulatory and professional bodies at a national level to accept learning outcomes and clinical practice competencies, and to incorporate them into suitable programs, is vital. Genomic education must be evidenced-based, flexible and responsive to changes in the education system and the workforce, provision of clinical services and conceptual and financial commitments to education. Genomic literacy cannot be static. The hallmark of the fast pace of genetic discovery is that "what is true today may not be true tomorrow."

Linking competencies with program accreditation and individual certification has been a major driver for the incorporation of genomics into the training and continuing education of the profession. It is essential that individuals and groups responsible for continuing education, curriculum development, licensing, certification, and accreditation of health professionals adopt the integration of genetics and genomic content into ongoing education. Although the diversity of speech-language pathology training models might suggest that no single genomics curriculum will work in all programs, it is incumbent upon speech-language pathology educators to leverage efforts to develop both a flexible core genetic curriculum based on appropriate competencies, as well as resources that can be adapted from different curricular structures.

It is crucial in the near future to implement and assess models of genomic education in speech-language pathology curricula and to measure whether they are effective in producing speech-language pathologists who are appropriately prepared to evaluate, manage, and support clients and families impacted by communication disorders, through their understanding of developments in genetics and genomics and the application of this knowledge to speech-language pathology.

Conclusions

Genomic education in the field of speech-language pathology is evolving, although slowly. Contributing to the pace and quality of genomic education are presuppositions, misconceptions, and misinformation constraining the effectiveness of the genetic and genomic education presently taking place at all levels of the continuum.

Genomic education must seek to explicitly address the discrepancies noted in this study and help close the gaps by facilitating the translation of current scientific knowledge. Conceptual change strategies must go steps further to address entrenched presuppositions, and misconceptions. These suppositions are fundamental to the representation and explanations of contemporary genomics, and constrain how effective the successful translation of genomics will occur in speech-language pathology.

Further information regarding attitudes, knowledge and needs are required from other stakeholders--students, instructors and faculty members, and individuals with communication disorders. In addition, determining the literacy levels of genomics in speech-language pathology of potential referral sources is a need. More referrals and collaborations with pediatricians, geneticists, genetic counselors, neurologists, and

otolaryngologists will occur as speech-language pathologists become increasingly genomic literate and competent.

Evidence-based, discipline-specific literacies, competencies and standards of practice will be required for the professional speech-language pathologist. Educators will be in need of measurement instruments of genomic literacy and competency, evidenced-based genomic curriculum models, and course design. Although the diversity of speech-language pathology training might suggest that no single genomic curriculum will work at all programs, it is incumbent upon speech-language pathology educators, administrators and policy makers to leverage efforts to develop a flexible core genetics and genomics curriculum founded on evidenced based competencies, as well as on core resources that can be adapted into different curricular structures at varying points along the education continuum. It is crucial in the near future to implement and assess models of genomic education in speech-language pathology curricula and to measure whether they are effective in producing (and maintaining through professional development events) speech-language pathologists who are appropriately prepared to evaluate, management and support communication sciences and its disorders in a genomic world. Based on current advances in genomics, providing a consistent level of genomic education should be our goal. It is hoped that the findings of the current study will provide a foundation for developing ways to facilitate genomic literacy and competency in speech-language pathology.

Discussion of the Research Process

Several limitations to the current study are identified and will inform further research.

Surveys

Representational Sample. The sample obtained from the population of 242 program directors in the United States appeared to be fairly representational on selected variables regarding degree programs: represented geographical areas, and types of degree programs offered. Population demographics on program directors were unavailable.

Sampling of the speech-language pathologists population in the United States began with random selection, however limited responses were received during the first week of survey solicitation. Attempts to increase the response rate led to non-random sampling techniques such as invitations on online community sites and snowball sampling. This resulted in a convenience sample, which limited the estimation of sampling error because the selection of responders was non-random. Convenience sampling may also result in exclusion bias. The results obtained in Survey II do appear to be representational of the national population, therefore information about the relationship between this sample and the population may be extrapolated, though with caution.

Response Rate. Program directors and professional speech-language pathologists in the United States were invited to participate in census surveys. With online surveys, cost issues did not limit the sample size, and all identifiable potential respondents who met inclusion criteria were invited to participate. The 29% response rate for the program directors correspond to rates of other surveys conducted with these populations. The response rate may have been enhanced by the population's anticipated interest in genomic literacy, and may have been restricted by timing. The survey was conducted in February and early March, when the half of the group with faculty positions may have had limited availability as new semesters in academic programs are beginning. In addition, it is possible that only those participants interested in genetic and genomics completed and returned the survey or likewise, only those who felt they had an understanding of genetics and genomics responded to the survey invitation. Inherent in all survey methodology is the possibility that participants' responses may differ from what they actually know or practice.

Survey Methodology and Design. Online survey methodology was well suited to the census survey, being convenient, cost efficient and versatile. Sending invitations via email allowed recruitment of a targeted population without incurring printing or postage costs. The SurveyMonkey.com platform supported both Likert-style questions for ranking concepts and text boxes to gather qualitative data. Displaying concepts related to each topical category on a single page allowed respondents to balance the relative importance of specific concepts.

During survey design, respondent burden was a concern due to the large number of items and sub-items. Two respondents made textual comments about the length of the survey. Five of 104 respondents failed to complete the entire Survey I, while 42

respondents did not complete the professional speech pathologist survey. Several items experienced a high skipped response on both surveys. Reasons for such may be related to the number of sub-items (for example, Item 16 in Survey II had 20 sub-items, with 56 skipped responses), the length and complexity of some of the items, and ambiguous questions. Review of the textual responses indicated ambiguity by three of the survey participants, and one respondent indicated that no definitions for the descriptors for the ratings were provided in the Likert scale.

Content Development. The first limitation of content development was that the size of the panel and the level of expertise of the panel which may be disputable. Locating a panel of judges who demonstrated a level of expertise in genetics, genomics and speech-language pathology was limiting. The panel members used in this study underwent an introduction to genomics in the field of speech-language pathology, which may have introduced investigator bias. A second limitation lies in the sample size for the focus groups and pilot studies. The number of participants in each focus group and pilot study for the surveys did not exceed eight. This is in part due to the limited availability of expert panel members.

A third limitation affecting content development was the use of constructs such as "awareness", "knowledge", and "confidence" within the domains of the surveys without providing a standard descriptor of the intended meanings. In addition, perceptions of what "*somewhat* confident" and "*significant* confidence" mean may vary across respondents.

Use of Core Competencies as a Framework. Development of the both surveys for each target group began with identifying an initial content domain for each instrument. This was a critical step, since the validity of the survey ultimately rests on the degree to which it can measure the attribute of interest. The challenge was to distill a large number of concepts embedded in a broadly-endorsed set of topics into a smaller number of concepts representing the current state of perceptions of genetics and genomics. The *Core Competencies* (NCHPEG, 2004, 2007) were used as a framework for several domains and items. However the Core Competencies provide broad guidelines with emphasis on principles of basic genetic science within realms of knowledge, skill and attitude. Discipline-specific constructs were not provided by the Core Competencies, but were extrapolated from benchmarks in the literature. Questions are raised as to the validity of using the *Core Competencies* as a primary framework for measuring compliancy.

Recognizing the rapid and escalating pace of genomic discovery, the Consensus Panel (2006) acknowledged the competencies were based on the "state of evidence available at the time they were developed" (p. 1). Since 2006, the bounds of genomic literacy have been strained by discovery of an increasing number of new phenomena, including epigenetic effects. Framing genomic literacy in conceptual understanding of fundamental concepts provides the best chance to prepare educators and clinical practitioners to implement genomic applications that cannot easily be predicted. Nevertheless, these concepts must be based on research-based evidence.

Speech-language pathologists who understand basic concepts of genetic structure, organization, and function will have a basis for understanding gene-based testing and

interventions. However, study results indicate that narrower guidelines accompanied by discipline specific genomic constructs, allowing for flexibility in application, are needed to establish genomic literacy and competency. This is true even as genomic discovery continues to mold basic conceptual understanding over time. Like the *Core Competencies* on which it is framed—and like the very definition of a gene itself—the ? the what? represents a work in progress. Not sure about this last sentence- does not seem to fit with the paragraph

While this investigation is a pioneer project, it is likely that others will contribute to the body of work providing empirical evidence for the applications and availability of genomic knowledge in the educational and clinical practices of speech-language pathology. Trust is placed in the knowledge that providing evidenced-based genomic education to both those in practice and those in training, and engaging them in the design of how this knowledge can be applied, will rapidly welcome the era of genomics to communication sciences and its disorders.

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Appendix A. IRB Approval

TEACHERS COLLEGE
COLUMBIA UNIVERSITY
OFFICE OF SPONSORED PROGRAMS

Institutional Review Board

February 7, 2012

Etoile LeBlanc
Teachers College
525 West 120th Street, Box 180
New York, NY 10027

Dear Etoile,

Thank you for submitting your study entitled, "*Needs Analysis of Genetic and Genomic in Communication Sciences: Implications for Change;*" the IRB has determined that your study is **Exempt** from review [Category **2**].

Please keep in mind that the IRB Committee must be contacted if there are any changes to your research protocol. The number assigned to your protocol is **12-153**. Feel free to contact the IRB Office [212-678-4105 or mbrooks@tc.edu] if you have any questions.

Please note that your consent form bears an official IRB authorization stamp. Copies of this form with the IRB stamp must be used for your research work.

Best wishes for your research work.

Sincerely,

Karen Froud, Ph.D.
Associate Professor of Speech and Language Pathology
Chair, IRB

cc: File, OSP

Appendix B. Survey I Cover Letter and Consent Form

TEACHERS COLLEGE
COLUMBIA UNIVERSITY

Survey I: Cover Letter and Consent Form

Curriculum Survey of Genetic Content of Speech Language Pathology Programs

Columbia University IRB Protocol Number: **12-153**

Dear Program Director,

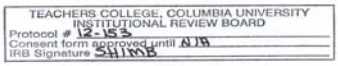
I am contacting you to invite you to participate in an online survey. The purpose of this study is to examine the presence and content of genetic and genomic content in the curriculum of academic degree program in speech language pathology. You will be contributing to knowledge about genetic education in speech language pathology.

You are asked to complete a survey, which will take 10 minutes to complete. The survey includes questions concerning; general demographic information about your institution and speech language pathology program, the presence and structure (delivery format and genetic genomic content) in the curriculum, perceptions, attitudes and opinions of genetics and genomics in regards to the role of speech language pathologist, and the perceived challenges faced by educators. The risk in participating in this study is minimal. There are not direct benefits from taking part in this study.

Your responses will be kept completely confidential. Your participation is voluntary. You will not be remunerated for your participation in this study. If you have concerns or questions about this study, please contact the IRB Office at 212-678-4105 or email at mbrooks@tc.edu. By completing the survey, you acknowledge that you have read this information and agree to participate in this research, with the knowledge that you are free to withdraw your participation at any time without penalty.

Thank you!

Etoile LeBlanc, MS.,CCC
Department of Biobehavioral Sciences
Program in Speech Language Pathology
Teachers College, Columbia University
New York, New York
eml2005@tc.columbia.edu



Appendix C. Survey I: Introductory Letter

TEACHERS COLLEGE
COLUMBIA UNIVERSITY

Pre Survey Introduction Letter

Using Genetics to Improve the Training of Speech Language Pathologists!

Columbia University IRB Protocol Number: **12-153**

February 2012

Dear Program Director,

I am writing to you to ask your assistance in gathering information on an issue affecting undergraduate and graduate degree programs in Speech Language Pathology. As you may be aware, there has been an explosion of genetic and genomic advancements in the last decade, which has a direct impact on communication sciences. However, it is suspected that the professional speech language pathologist, and the training of speech language pathologist are ill prepared to effectively use genetic and genomics in clinical and research practices.

I have designed a questionnaire assessing the presence and the extent of genetic and genomic content in speech language pathology degree programs. This survey called, *Genetic Content in the Curriculum of Speech Language Pathology Degree Programs* should take you only 10-15 minutes to complete.

Your participation would provide invaluable information on how we can best improve the education and training of speech language pathologists. I welcome any questions you may have. I can be reached at craniofacial@msn.com.

Thank you in advance for contributing to the improvement of the field of communication sciences. You will be receiving an email in the next several days, inviting you to participate in this survey. I trust you will take the time to participate in the opportunity to contribute.

Thank you!

Etoile LeBlanc, MS.,CCC
Department of Biobehavioral Sciences
Program in Speech Language Pathology
Teachers College, Columbia University
New York, New York

Appendix D. Genomic Education Rubric

Development of a Model of Genomic Education

Summative evidence on whether training programs were including genetic and genomic content in their curricula is salient, however formative attributes of genetic and genomic content provide the qualitative evidence on genomic literacy and competency of the student. A rubric format best provides formative, efficient, and transparent feedback for rating thematic concepts. It also provides a framework helpful to curriculum development, student learning, and professional development.

The rubric system was developed to act as a model of genomic education, from which comparisons can be formulated. The rubric's framework is loosely based on theories of science education literature (Eijk, 2010). Science literacy is defined in terms of what it means to *know* and to *learn* in addition specific constructs of genetic and genomic science. Inherent in this framework is that genomic science education is dynamic, increasingly reflexive (availability of sophisticated data easily accessible by the Internet in very short periods of time), and possesses transdisciplinarity (having the ability to bring together different genomic disciplines to generate new meaning from huge data sets), ultimately generating new disciplines further fostering scientific literacy.

The information will provide a qualitative measure of genetic and genomic education in degree programs. The rubric is based on thematic concepts across academic and clinical need of a Masters level student in speech language pathology, and constructs of knowledge and pedagogy based on a constructionists perspective. The following thematic concepts are all integral attributes of genomic education: 1) type of genetics and genomics, 2) type of content, 3) model of knowledge, 4) pedagogic model, and 5) the level of expected literacy and competency of the students. The rubric is the first step in operationally defining constructs necessary for genomic education in speech language pathology.

Establishing the rubric as based on three phases of development: Phase 1) determining a foundational content and level of genetic and genomic principles from which a degree of literacy and competency can be expected from students, Phase 2) providing content validity, and Phase 3) developing a means to assess these rubric thematic concepts within the survey.

Phase 1

To determine formative information of genetic and genomic constructs in curricular content, thematic concepts were developed. These were based on a) benchmarks in the literature, b) ASHA competencies and practice standards (ASHA, Standard IIIB, 2007) curriculum requirements of the CAPCSD (2012), and Core Competencies (NCHPEG, 2007), and c) Roger's knowledge framework (2003) and Ausubel's, theory of learning and assimilation (Woolfolk, Winne, Perry, & Skapka, 2010). What follows is a description of the development of the model and the manner of its use to determine summative and formative evidence of genetic content in training programs.

Rubrics of Genomic Education

Five foundational conceptual thematics were delineated to represent a rubric: 1) general genetic and genomic knowledge, 2) discipline-specific genomic knowledge, 3) type of pedagogic model used (genetic or genomic), 4) level of theoretical knowledge (curriculum format and content reflecting basic conceptual "awareness" rather than task-oriented "how-to" knowledge), 5) type and level of learning model-literacy and competency construct.

1) General Genetic and Genomic Knowledge. In the second edition of the Core Competencies, a consensus panel (2007) identified broad areas of knowledge along with clinical performance or practice indicators (as indicated as "skill" and "attitudes") for each competency. To narrow the width of the competencies, further refinement of

thematic concepts most foundational to genomic literacy and competency in curricular content needed to be established. While the Core Competencies provide a broad benchmark, they were written as literacy and competencies for which a set of foundational concepts had not been clearly explicated for the speech language pathologist's specific work practices, let alone for the student. Furthermore, the breadth of knowledge subsumed by the Core Competencies exceeds the scope of a concept inventory (i.e., cytogenetics, molecular sciences) for the speech language pathologist. Therefore, identification was needed of genetic and genomic concepts that are (a) aligned with the Core Competencies, and (b) are most salient to speech language pathology training programs.

To begin, the Core Competencies were deconstructed to identify embedded concepts. The list of concepts was prioritized according to relevance to speech language pathology practice at the Masters level and reduced to include the most important concepts. Both the knowledge areas and the clinical performance indicators were deconstructed to identify supporting genetic and genomic concepts, i.e., basic foundational knowledge required to achieve each competency. Wide variance was evident in the knowledge necessary to achieve various competencies. Certain professional responsibility competencies require little understanding specific to genetics or genomics, while various competencies in the professional practice domain necessitate understanding of multiple concepts. The Core Competencies identified specific areas of knowledge along with clinical performance or practice indicators (delineated as "skill" in the Core Competencies). Certain concepts, such as the use of family history, were embedded in more than one competency. What resulted, 29 extracted concepts from the Core Competencies, representing basic principles in genetic and genomic science, used to set the foundation for discipline-specific knowledge.

2) Discipline-specific Genetic and Genomic Knowledge. Eighteen concepts were established from the method used for extracting general genetic and genomic knowledge

with the exception of also using benchmarks in the literature to reflect discipline specific knowledge constructs such as, representing discipline specific as well as concepts specific to speech language pathology (i.e., the relevancy of certain genes such as FOXP2).

3) Genetic/Genomic Model. The Human Genome Project and advancements of genetic, epigenetic and genomic information has challenged and changed the field of genetics. The field has been classified into two primary fields; genetics versus genomics, each representing two eras; pre-genomics versus post-genomics. Historically, genetic instruction emphasized Mendelian ratios (certain pattern of inheritance) and monogenic traits and disorders. Additive to this perspective was the "introduction" to the unknown contributions of gene-environmental interactions. Yet, much of the focus in teaching genetics had been on classical genetics, an outdated perspective, often perpetuating misconceptions (Dougherty, 2009). Modern genetics, also known as post-genomic genetics, has a much more advanced understanding of the role of the environment at various levels of complexity, giving way to the polygenic expression of all human traits, diseases, and disorder modulated by many complex environments.

The rubric was designed to top reflect the type of genetic and genomic information being shared. Discerning the type or model used in curricula elucidates the level (state-of-the-art) of information is being shared with the students, may shed information on the instructors understanding of the field, and will demonstrate areas of further research.

4) Level of Literacy and Competency. There is a difference between the teaching model of the level of model used or what is expected) and the level of literacy and competency demonstrated by the student. The current rubric Literacy is more closely aligned with knowledge, while competency infers the ability to apply that knowledge.

5) Level of Knowledge. There are many possible frameworks for organizing components of professional literacy and competency. One might separate knowledge from skill or view skill as demonstration of knowledge. For example, Bloom's taxonomy

of educational objectives in the cognitive domain illustrates depth of knowledge. Lower levels include knowledge of facts and methods and higher levels of understanding involve synthesis and evaluation of facts and methods. E.M. Rogers's (2003) framework of knowledge works on the same premise yet as a process rather than a linear delineation. Conceptual knowledge pertains to theories, ideas and factual information one has stored in memory, and is typically learned in academic instruction and is similar to Bloom's lower level of taxonomy. Process knowledge of know-how (Rogers, 2003) is an essentially meta-cognitive type of knowledge and includes a skill- accomplishing a task. The mission of graduate programs is to provide a pedagogic environment of both conceptual and process knowledge.

Phase 2: Content Validity of the Thematic Rubric

The rubrics was presented to a focus group, comprised of a geneticist, a faculty member of a graduate program in speech language pathology, and two speech language pathologists, with particular expertise in craniofacial speech language disorders. The focus group was asked to respond to the concept list by rating (categorical value +/-) each subconstruct of each item according to the thematic concepts presented in the rubric. The mean value was calculated for each concept.

Phase 3: Measurement of Thematic Concepts in the Curriculum

It is difficult to assess the quality of the genetic content by means of an online survey whose responses are given by someone who is not the individual teaching the targeted course. However, one may assess the thematic concepts through the item responses across programs and make inferences on summative and formative data. Content analysis is a method of objective, systematic and quantitative description of manifest and latent content of communication (Graneheim & Lundman, 2004). Content analysis enables a researcher to systematically and efficiently identify trends or patterns (Stemler, 2001). Content analysis for the purposes of this needs assessment was mapped

to specific thematic concepts. For each of the 29 subconcepts, representing 4 rubric concepts, the program directors responses were converted to categorical values (+/-).

Responses from the survey were analyzed using content analysis, applying the method of Krippendorff (1980) and a procedure described by Garvin-Doxas and Klymkowsky (2007). Rubric constructs were mapped to survey items 7, 16, and 17. While these mapped concepts may not represent the only genetic concepts offered in the curriculum, they are judged to be adequate markers for qualifying genetic and genomic content.

Appendix E. Content Validity for Genomic Education Rubric

Items	Response Given by Ratio (Scale/Subscale)																Content Validity Index (CVI)
	Judge 1				Judge 2				Judge 3				Judge 4				
Competencies expected from graduated students	1/4		3/4	4/3	1/4	2/4	3/4	4/3	1/4	2/4	3/2	4/4	1/2	2/4	3/4	4/4	.843
Courses in which genetic and genomic content are integrated	1/2	2/2	3/3	4/4	1/3	2/4	3/3	4/4	1/4	2/2	3/4	4/3	1/4	2/4	3/4	4/4	.812
Genetic and genomic topics integrated into content	1/4	2/4	3/3	4/3	1/4	2/4	3/4	4/4	1/2	2/4	3/2	4/2	1/4	2/3	3/2	4/2	.796
Content Validity Index for Genomic Education Rubric																	.817

Appendix F. Survey I: Focus Group Results and Comments

Focus Group Responses for Survey I	Frequency N=8		Frequency N=8		
	Does this question test the aim		Is this a quality question?		Text Comments
	Yes	No	Yes	No	
1. Does your program offer a degree program in speech-language pathology?	7	1	8	0	
2. In what State is your program located?	8	0	8	0	
3. How long has your institution been offering a program in speech language pathology?	8	0	8	0	
4. What degree(s) does your program offer in speech-language pathology?	8	0	8	0	Add- "Check all that apply"
5. Is your speech-language pathology program affiliated with a medical school?	7	1	8	0	Add – "and or a teaching hospital"
6. Is your speech-language pathology program designated as medically-based?	7	1			
7. The following is a list of knowledge, skills, and attitudes often required to be achieved by graduated speech-language pathologists. Please check all competencies your speech-language pathology program expects your students should demonstrate upon graduation.	6	2	8	1	Too many subscales
8. Does your speech-language pathology program include genetic content in your speech-language pathology curriculum?	8	0	8	0	
9. What may be the reasons why genetic/genomic content is not being offered ?	8	0	8	0	Add- "Check all that apply"
10. Are there plans to develop genetic and genomic content in the curriculum within the next two years?	6	2	7	1	Change to 5 years
11. Describe the structure of genetic content offered in the curriculum.	8	0	8	1	Consider asking the percent of genetic content
12. How often is the course offered?	8	0	8	0	
13. Is this a required or an elective course?	8	0	8	0	Is this question necessary?
14. If the course is an elective course, approximately what percent of students are enrolled in the class each time it is offered?	7	1	7	1	
15. In what year of the student's degree program is a course(s) with genetic and or genomic content typically taken?	8	0	8	0	Why is this question asked?
16. In which course(s) is genetic/genomic content integrated?	8	0	8	0	
17. What topics of genetics/genomics are covered in your curriculum?	8	0	8	0	
18. Describe the characteristics of the instructor teaching the course with genetic/genomic content?	8	0	8	0	Add- "is not a requirement" Add-in if accreditation issue

19. What type of instructional method is typically used in teaching the genetics/genomics course(s). Check all that apply.	8	0	8	0	
20. In your opinion, how much interest in genetics/genomics is generated by your students?	7	1	7	1	
21. In your opinion, how much interest in genetics/genomics is generated by your faculty?	7	1	7	1	
22. Does your department engage in genetically based speech-language pathology research?	7	1	8	0	
23. What area(s) of research is currently being conducted?	7	1	8	0	
24. In your opinion, do you believe the graduating speech language pathology student is prepared with the appropriate knowledge and skills in genetics and genomics?	8	0	8	0	
25. In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?	8	0	8	0	
26. With continued advances in genetics and or genomics, do you think the impact on clinical practices of the speech-language pathologist will change in the next 5-10 years?	8	0	8	0	
27. Do you believe that genetics and genomics as a role in the clinical practice of speech-language pathology?	8	0	8	0	
28. Of the following areas of study, which ones doe you believe genetics and genomics has a role?	8	0	8	0	
29. How long have you held your position in the speech-language pathology department?	8	0	8	0	
30. How many faculty are on staff?	3	5	2	6	Has no relevance
31. Are you the program director of the speech-language degree program?	8	0	8	0	
32. What is the primary position in the speech-language pathology program?	8	0	8	0	Change “the” to “your”
33. In what area are you trained?	8	0	7	1	Re-word the question
34. Have you attended educational events with genetic and genomic content?	2	6	1	8	Not relevant
35. What is your opinion of Genetic Information Non-discrimination Act of 2008?	0	8	7	1	Assumes knowledge; not relevant to aims
36. Have you taught a craniofacial and or cleft palate course?	1	7	0	8	Leading question
37. How much genetic content was included in the course?	1	7	0	8	Not appropriate
38. How much genomic content was included in the course?	1	7	0	8	Not appropriate
39. Were you satisfied in the amount of genetic and genomic content in the course?	0	8	0	8	Not appropriate question
40. In your opinion, do you believe ASHA should mandate policies regarding competencies in genetic and genomic related skills?	1	7	2	6	Leading question; nice to know but a lot of assumptions being made
41. In your opinion, do you believe degree programs should be required to provide genetic and genomic content in the curriculum?	0	8	3	5	Leading question
42. Do you believe there is a genetic relationship to	0	8	0	8	Not part of the aim of

apraxia?					the study
43. Do you believe there is a genetic relationship to autism?	0	8	0	8	Same as above
44. Do you believe there is a genetic relationship to language disorders?	0	8	0	8	Same as above
45. Do you believe speech-language disorders are inherited?	0	8	0	8	Same of above
46. Do you believe there is a genetic relationship to stuttering?	0	8	0	8	Same as above
47. Do you believe there is a genetic relationship to voice disorders?	0	8	0	8	Same as above

Appendix G. Survey I: Content Validity Results of Pilot Study

Survey I Questions* Subscales of each item can be viewed in Appendix	Response Given by Ratio (Scale/Subscale)																Content Validity Index (CVI)
	Judge 1				Judge 2				Judge 3				Judge 4				
1. Does your program offer a degree program in speech-language pathology?	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/4	.968
2. In what State is your program located?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
3. How long has your institution been offering a program in speech language pathology?	1/4	2/4	3/3	4/3	1/4	2/4	3/4	4/4	1/2	2/4	3/2	4/2	1/4	2/2	3/3	4/2	.796
4. What degree(s) does your program offer in speech-language pathology?or genomic related?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
5. Is your speech-language pathology program affiliated with a medical school?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
6. Is your speech-language pathology program designated as medically-based?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
7. The following is a list of knowledge, skills, and attitudes often required to be achieved by graduated speech-language pathologists.	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00

8. Does your speech-language pathology program include genetic content in your speech-language pathology curriculum?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
9. What may be the reasons why genetic/genomic content is not being offered ?.	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/3	3/4	4/4	.953
10. Are there plans to develop genetic and genomic content in the curriculum within the next two years?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
11. Describe the structure of genetic content offered in the curriculum.	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/4	1/4	2/3	3/3	4/3	1/4	2/4	3/4	4/4	.921
12. How often is the course offered?	1/4	2/2	3/2	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/2	3/2	4/3	.765
13. Is this a required or an elective course?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
14. If the course is an elective course, approximately what percent of students are enrolled in the class each time it is offered?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
15. In what year of the student's degree program is a course(s) with genetic and or genomic content typically taken?	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/4	.968
16. In which course(s) is genetic/genomic content integrated?	1/4	2/3	3/3	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/2	3/3	4/4	.906

17. What topics of genetics/genomics are covered in your curriculum?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
18. Describe the characteristics of the instructor teaching the course with genetic/genomic content?	1/1	2/4	3/3	4/4	1/3	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.875
19. What type of instructional method is typically used in teaching the genetics/genomics course(s). Check all that apply.	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
20. In your opinion, how much interest in genetics/genomics is generated by your students?	1/3	2/4	3/4	4/4	1/2	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	9.53
21. In your opinion, how much interest in genetics/genomics is generated by your faculty?	1/2	2/4	3/4	4/4	1/1	2/4	3/3	4/4	1/3	2/3	3/4	4/4	1/3	2/4	3/4	4/4	8.53
22. Does your department engage in genetically based speech-language pathology research?	1/1	2/4	3/4	4/3	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	8.43
23. What area(s) of research is currently being conducted?	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/3	8.90
24. In your opinion, do you believe the graduating speech language pathology student is prepared with the appropriate knowledge and skills in genetics and genomics?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00

25. In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
26. With continued advances in genetics and or genomics, do you think the impact on clinical practices of the speech-language pathologist will change in the next 5-10 years?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
27. Do you believe that genetics and genomics as a role in the clinical practice of speech-language pathology?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
28. Of the following areas of study, which ones doe you believe genetics and genomics has a role?	1/3	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.973
29. How long have you held your position in the speech-language pathology department?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
30. Are you the program director of the speech-language degree program?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00

31. What is the primary position in the speech-language pathology program?	1/4	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
32. In what area are you trained?	1/4	2/2	3/2	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/2	3/2	4/3	.765

Appendix H. Survey I: Text Responses Provided by Program Directors

Question 7

The following is a list of knowledge, skills, and attitudes often achieved by graduating speech language pathologists.
1. "Students will have foundational knowledge that will allow them to recognize factors and counsel to the extent of referral for more services as needed."
2. "Some of these would be carried out somewhat superficially, with referral to someone who could do them more in-depth fashion."
3. "I am not sure of intent but we typically refer to UCHSC (e.g., childrens hospital/UH) when we suspect the family is in need of genetic services and more precise diagnosis and counseling."
4. "We teach students to counsel as related to communication disorders only- not medical or genetic counseling. This is not within our scope of practice."

Question 10.

Are there plans to develop a genetic and genomic content in the curriculum within the next five years?
1. "But we may consider it.'
2. "It would be difficult to fit into students Plan of Study for completion of Masters in 6 semesters and would need to hire additional faculty member."
3. "Faculty is looking at complete overhaul of curriculum within the next two years, and likely new areas will be considered at that time."
4. "Perhaps."

Question 16

In which course(s) is genetic and genomic content offered?
1. " It is a combined course with Autism and AAC."
2. "Fluency" X2
3. "Small amounts in other courses"
4. "Counseling"

Question 18

Describe the characteristics of the instructor teaching with course with genetic/genomic content? Check all that apply.
1. "Team taught by Audiologists and Nurse- both with interest and study in genetics."
2. "Guest lectures"
3. "with much craniofacial experience and with courses in genetics
4. "instructor completed coursework in genetic counseling as part of doctoral degree'

Question 19

What type of instructional method is typically used in teaching the genetics/genomics course(s)? Check all that apply.
1. "Web-based module."
2. "Genetics and Syndromes elective has been taught as traditional class and on-line distance Ed class."
3. "Problem-based group projects."

Question 20

"In your opinion how much interest in genetics/genomics is generated by your students.
1. "There were 4 questions on the PRAXIS this year which they took before they did any genetics except for what they did at undergrad level."
2 Very little… they groan all the way through it."

Question 21

"In your opinion how much interest in genetics/genomics is generated by your faculty.
1. "under discussion."

Question 22.

Does your department engage in genetically-based speech-language pathology research?
1. "Not to any large extent. One researcher."
2. "Instructor has completed research in genetics education for non-geneticist research."

Question 24.

In your opinion, do you believe the graduating speech-language pathology student is prepared with the appropriate knowledge and skills in genetics and genomics?
1. "Foundational but no in-depth knowledge."
2. "As with any topic, we would like to provide more training that we have time for."
3. "We graduate beginning clinicians. I expect them to continue learning in many topics."
4. "not clear on what we should be teaching them at this point, but definitely something to consider more"
5. There is always room for more. It is impossible to include everything in a Masters level graduate program. Students specialize upon graduation."
6. Knows enough to be part of the conversation and treatment and knows when to refer."

Question 25.

In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?
1. "My response is based on a consideration of genetics relative to the entire scope of clinical problems."
2. "Do you mean how much do we take into account – far less than we should. Do you mean how important it is –significantly so."
3. "Presently minimal but this will change."
4. "Not enough information is filtering down to the practitioner"
5. "Genetics certainly plays a role in dx and tx, but "the current field" is not changing my practice much."

Question 26.

With continued advances in genetics and genomics, do you think the impact on clinical practices of the speech language pathologist will change in the next 5-10 years?
1. "Probably greater changes in the 10-20 yr range."

Question 27.

Do you believe genetics/genomics has a role in the clinical practice of speech language pathology?
1. "depends on the setting."
2. "But not a major one."
3. "However, I do not feel we should play the roles of genetic diagnosticians or genetic counselor. There are people who specialize in those areas and we should refer to them."
4. "Not enough information is filtering down to the practitioner"
5. "Considering our scope, minimally so, but yes."

Appendix I. Survey II: Cover Letter and Consent Form

TEACHERS COLLEGE
COLUMBIA UNIVERSITY

Survey II: Cover Letter and Consent Form

Genetics in the Post-Genomic Era: A Practitioner's Perspective

Columbia University IRB Protocol Number: **12-153**

Dear Speech Pathologist,

I am contacting you to invite you to participate in an online survey. The purpose of this study is to examine the how genetics and genomics impact clinical practices of speech language pathologists. You will be contributing to knowledge about genetic education in speech language pathology.

You are asked to complete a survey, which will take 10-15 minutes to complete. The survey includes questions concerning; assessing your opinions, work practices, and perceived knowledge of genetics and genomics. There are not direct benefits from taking part in this study.

Your responses will be kept completely confidential. Your participation is voluntary. You will not be remunerated for your participation in this study. If you have concerns or questions about this study, please contact the IRB Office at 212-678-4105 or email at mbrooks@tc.edu.

By completing the survey, you acknowledge that you have read this information and agree to participate in this research, with the knowledge that you are free to withdraw your participation at any time without penalty.

Thank-you!

Etoile LeBlanc, MS.,CCC
Department of Biobehavioral Sciences
Program in Speech Language Pathology
Teachers College, Columbia University
New York, New York
eml2005@tc.columbia.edu

TEACHERS COLLEGE
COLUMBIA UNIVERSITY

Survey II: Pre-Survey Introduction Letter

Genetics in the Post-Genomic Era: A Practitioner's Perspective

Columbia University IRB Protocol Number: **12-153**

February 2012

Dear Program Director,

I am writing to you to ask your assistance in gathering information on your perceptions of genetics and genomics and its role in speech language pathology. As you may be aware, there has been an explosion of genetic and genomic advancements in the last decade, which has a direct impact on communication sciences. However, it is suspected that the professional speech language pathologist may feel ill prepared to effectively understand or use genetic and genomic advancements in clinical practices.

I have designed a questionnaire assessing your opinions, work practices, and perceived knowledge of genetics and genomics. This survey called *Genetics in the Post-Genomic Era:A Practitioner's Perspective* should take you only 10-15 minutes to complete. There are not direct benefits from taking part in this study.

Your participation would provide invaluable information on how we can best improve the education and training of the members of communication sciences. I welcome any questions you may have. I can be reached at eml2005@tc.columbia.edu.

Thank you in advance for contributing to the improvement of the field of communication sciences. You will be receiving an email in the next several days, inviting you to participate in this survey. I trust you will take the time to participate in the opportunity to contribute.

Thank you!

Etoile LeBlanc, MS.,CCC
Department of Biobehavioral Sciences
Program in Speech Language Pathology
Teachers College, Columbia University
New York, New York
eml2005@tc.columbia.edu

Appendix J. List of ASHA Special Interest Groups

SIG 1. Language Learning and Education
SIG 2. Neurophysiology and Neurogenic Speech and Language Disorders
SIG 3. Voice and Voice Disorders
SIG 4. Fluency and Fluency Disorders
SIG 5. Speech Science and Orofacial Disorders
SIG 6. Hearing and Hearing Disorders: Research and Diagnostics
SIG 7. Aural Rehabilitation and Its Instrumentation
SIG 8. Public Health Issues Related to Hearing and Balance
SIG 9. Hearing and Hearing Disorders in Childhood
SIG 10. Issues in Higher Education
SIG 11. Administration and Supervision
SIG 12. Augmentative and Alternative Communication
SIG 13. Swallowing and Swallowing Disorders (Dysphagia)
SIG 14. Communication Disorders and Sciences in Culturally and Linguistically Diverse (CLD) Populations
SIG 15. Gerontology
SIG 16. School-Based Issues
SIG 17. Global Issues in Communication Sciences and Related Disorders
SIG 18. Telepractice

Appendix K. Survey II: Introductory Letter

TEACHERS COLLEGE
COLUMBIA UNIVERSITY

Survey II: Pre-Survey Introduction Letter

Genetics in the Post-Genomic Era: A Practitioner's Perspective

Columbia University IRB Protocol Number: **12-153**

February 2012

Dear Program Director,

I am writing to you to ask your assistance in gathering information on your perceptions of genetics and genomics and its role in speech language pathology. As you may be aware, there has been an explosion of genetic and genomic advancements in the last decade, which has a direct impact on communication sciences. However, it is suspected that the professional speech language pathologist may feel ill prepared to effectively understand or use genetic and genomic advancements in clinical practices.

I have designed a questionnaire assessing your opinions, work practices, and perceived knowledge of genetics and genomics. This survey called *Genetics in the Post-Genomic Era:A Practitioner's Perspective* should take you only 10-15 minutes to complete. There are not direct benefits from taking part in this study.

Your participation would provide invaluable information on how we can best improve the education and training of the members of communication sciences. I welcome any questions you may have. I can be reached at eml2005@tc.columbia.edu.

Thank you in advance for contributing to the improvement of the field of communication sciences. You will be receiving an email in the next several days, inviting you to participate in this survey. I trust you will take the time to participate in the opportunity to contribute.

Thank you!

Etoile LeBlanc, MS.,CCC
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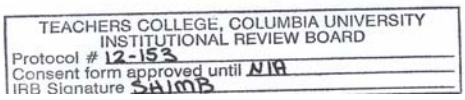


Appendix L. Survey II: Focus Group Results and Comments

Focus Group Responses for Survey II	Frequency N=8		Frequency N=8		
	Does this question test the aim?		Is this a quality question?		Text Comments
	Yes	No	Yes	No	
1. Are you aware of any advances in genetics/genomics within the last 5-10 years that directly related to speech-language pathology?	7	1	8	0	More info from TV and NY Times than ASHA
2. In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?	8	0	8	0	
3. Have you had any formal educational course(s) in any topic of genetics. If you have formal education, fill out the year you completed such course(s)?	6	2	7	1	Will all be able to remember dates and content?
4. Have you attended any ASHA sponsored continuing education events where the content was genetic or genomic related?	8	0	8	0	Same as above
5. What year(s) did you attend an ASHA CEU event in which genetics and genomics was included in the content of the event. Check all that apply.	7	1	8	0	Scale is a little confusing
6. In your opinion, how well did your speech-language pathology degree program prepare you to understand the current field of genetics and genomics?	7	1	8	0	
7. Do you feel confident in your understanding of the current field of genetics and genomics as it applies to the field of speech-language pathology?	6	2	8	1	
8. Of the following, which area(s) of communication sciences do you believe genetics and or genomics plays a role? Check all that apply?	8	0	8	0	
9. Have you received questions from parents/ patients/clients regarding genetic and genomic principles of basic science such as ways in which conditions or traits are inherited, risks for recurrence of a condition or trait, etc.	8	0	8	0	
10. Have you received questions from parents/patients/clients and speech-language pathology?	6	2	7	1	
11. Rate your level of confidence in knowing you were providing the appropriate genetic/genomic information/answers to their questions.	8	0	8	1	
12. Of the following activities involved in the assessment and management of speech-language development and disorders, indicate which activity(ies) you have or are currently performing and the level of confidence you feel you posses for each activity?	6	2	8	0	Too many subscales
13. How confident are you in providing counseling/information on genetic/genomic speech-language pathology issues?	8	0	8	0	
14. Do you feel confident in recognizing a speech-language disorder that may have a genetic or genomic relationship?	7	1	7	1	Is this question necessary?
15. To which professional would you first refer, if you suspected a genetic/genomic relationship occurring in patient/client? Check all that apply.	8	0	8	0	Why is this question asked?

16. Do you feel you have the knowledge to answer the following questions?	8	0	8	0	doubt many will have the knowledge
17. How long have you been practicing as a speech-language pathologist?	8	0	8	0	
18. What is your age?	5	3	5	3	Relevance?
19. Are you male or female?	8	0	8	0	
20. In what year did you complete your most advanced degree?	7	1	7	1	
21. What degree(s) do you currently hold. Check all that apply?	7	1	7	1	
22. Are you currently completing a degree?	7	1	8	0	
23. What degree are you currently completing and in what year?	7	1	8	0	
24. What is your current certification status?	8	0	8	0	
25. Are you currently engaged in clinical practice?	8	0	8	0	
26. What is/are your specialty area(s) of practice? Check all that apply.	8	0	8	0	
27. What is your primary work setting? Check only one.	8	0	8	0	
28. Are you teaching in the field of speech-language pathology?	8	0	8	0	
29. What academic level and what course(s) do you teach?	8	0	8	0	
30. Are you currently conducting research that involves genetics or genomics?	3	5	2	6	Has no relevance to the clinician
31. Are you currently or have been in the past a member of a craniofacial and or cleft palate team/center?	8	0	8	0	
32. In what city and state is/was the team or center located?	8	0	8	0	What is the relevance?
33. Was a geneticist or genetic counselor present at the team meetings at least 80% of the team?	8	0	7	1	Re-word the question
34. How would you rate your level of confidence in understanding genetics and its recent advances within your role as a speech-language pathologist on the team?	6	2	7	1	

Appendix M. Survey II: Content Validity Results of Pilot Study

Survey II Questions	Response Given by Ratio (Scale/Subscale)																Content Validity Index (CVI)
	Judge 1				Judge 2				Judge 3				Judge 4				
1. Are you aware of any advances in genetics/genomics within the last 5-10 years that directly related to speech-language pathology?	1/3	2/4	3/3	4/4	1/4	2/3	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/4	.936
2. In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?	1/4	2/4	3/3	4/3	1/4	2/4	4/3	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.953
3. Have you had any formal educational course(s) in any topic of genetics. If you have formal education, fill out the year you completed such course(s)?	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/4	1/2	2/4	3/2	4/2	1/4	2/2	3/3	4/2	.781
4. Have you attended any ASHA sponsored continuing education events where the content was genetic or genomic related?	1/4	2/2	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/2	3/4	4/4	1/4	2/3	3/4	4/4	.922

5. What year(s) did you attend an ASHA CEU event in which genetics and genomics was included in the content of the event. Check all that apply.	1/2	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/2	3/4	4/4	.937
6. In your opinion, how well did your speech-language pathology degree program prepare you to understand the current field of genetics and genomics?	1/2	2/2	3/4	4/4	1/3	2/4	3/3	4/4	1/3	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.890
7. Do you feel confident in your understanding of the current field of genetics and genomics.	1/2	2/3	3/3	4/3	1/4	2/4	3/4	4/4	1/3	2/4	3/3	4/4	1/3	2/4	3/4	4/4	.875
8. Of the following, which area(s) of communication sciences do you believe genetics and or genomics plays a role? Check all that apply?	1/4	2/4	3/4	4/3	1/3	2/3	3/3	4/3	1/3	2/4	3/4	4/4	1/3	2/3	3/3	4/3	.843
9. Have you received questions from parents/ patients/clients regarding genetic and genomic principles of basic science such as ways in which conditions or traits are inherited, risks for recurrence of a condition or trait, etc.	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/3	3/4	4/4	.953

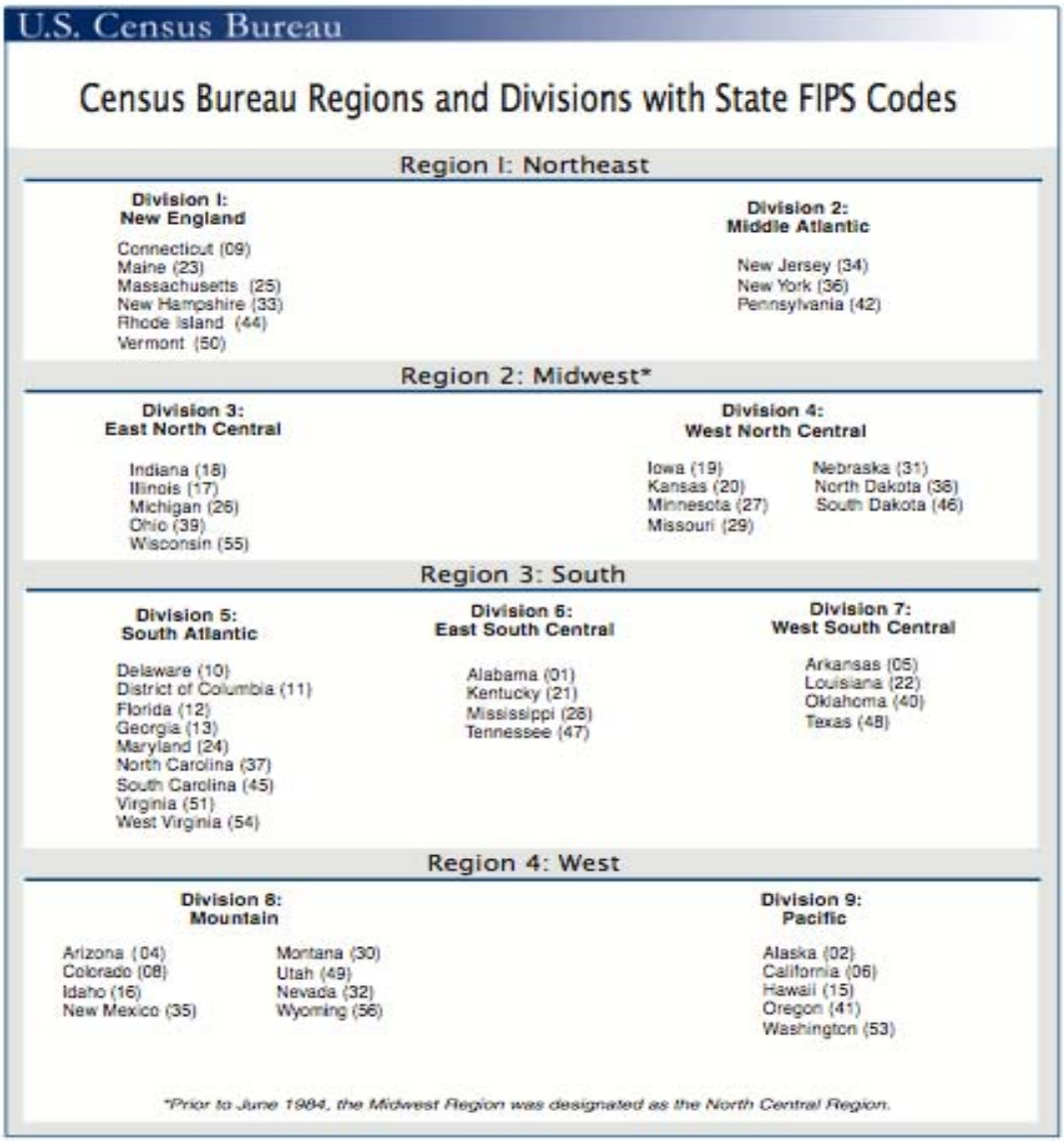
10. Have you received questions from parents/patients/clients and speech-language pathology?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
11. Rate your level of confidence in knowing you were providing the appropriate genetic/genomic information/answers to their questions.	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/4	1/4	2/3	3/3	4/3	1/4	2/4	3/4	4/4	.921
12. Of the following activities involved in the assessment and management of speech-language development and disorders, indicate which activity(ies) you have or are currently performing and the level of confidence you feel you posses for each activity?	1/4	2/2	3/2	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/2	3/2	4/3	.765
13. How confident are you in providing counseling/information on genetic/genomic speech-language pathology issues?	1/3	2/3	3/3	4/3	1/3	2/3	3/3	4/3	1/3	2/4	3/4	4/4	1/3	2/4	3/4	4/3	.828
14. Do you feel confident in recognizing a speech-language disorder that may have a genetic or genomic relationship?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.843

15. To which professional would you first refer, if you suspected a genetic/genomic relationship occurring in patient/client? Check all that apply.	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/4	.968
16. Do you feel you have the knowledge to answer the following questions?	1/4	2/3	3/3	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/2	3/3	4/4	.906
17. How long have you been practicing as a speech-language pathologist?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
18. What is your age?	1/1	2/4	3/3	4/4	1/3	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.875
19. Are you male or female?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
20. In what year did you complete your most advanced degree?	1/4	2/3	3/3	4/3	1/2	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/3	.860
21. What degree(s) do you currently hold. Check all that apply?	1/2	2/4	3/4	4/4	1/1	2/4	3/3	4/4	1/3	2/3	3/4	4/4	1/3	2/4	3/4	4/4	8.53
22. Are you currently completing a degree?	1/1	2/4	3/4	4/3	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	8.43
23. What degree are you currently completing and in what year?	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/3	8.90
24. What is your current certification status?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
25. Are you currently engaged in clinical practice?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00

26. What is/are your specialty area(s) of practice? Check all that apply.	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
27. What is your primary work setting? Check only one.	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
28. Are you teaching in the field of speech-language pathology?	1/3	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	.973
29. What academic level and what course(s) do you teach?	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
30. Are you currently conducting research that involves genetics or genomics?	1/3	2/4	3/3	4/4	1/4	2/4	3/4	4/3	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/3	.973
31. Are you currently or have been in the past a member of a craniofacial and or cleft palate team/center?	1/4	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/1	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1.00
32. In what city and state is/was the team or center located?	1/4	2/3	3/4	4/1	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/4	2/3	3/2	4/4	.765
33. Was a geneticist or genetic counselor present at the team meetings at least 80% of the team?	1/4	2/4	3/3	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/4	4/4	1/4	2/4	3/3	4/4	.875

34. How would you rate your level of confidence in understanding genetics and its recent advances within your role as a speech-language pathologist on the team?	1/4	2/3	3/3	4/4	1/4	2/4	3/3	4/1	1/4	2/4	3/4	4/4	1/3	2/2	3/3	4/3	.750

Appendix N. US Geographical Areas as Per the US Census Bureau



U.S. Census Bureau

Census Bureau Regions and Divisions with State FIPS Codes

Region I: Northeast

Division I: New England

Connecticut (09)
Maine (23)
Massachusetts (25)
New Hampshire (33)
Rhode Island (44)
Vermont (50)

Division 2: Middle Atlantic

New Jersey (34)
New York (36)
Pennsylvania (42)

Region 2: Midwest*

Division 3: East North Central

Indiana (18)
Illinois (17)
Michigan (26)
Ohio (39)
Wisconsin (55)

Division 4: West North Central

Iowa (19)
Kansas (20)
Minnesota (27)
Missouri (29)
Nebraska (31)
North Dakota (38)
South Dakota (46)

Region 3: South

Division 5: South Atlantic

Delaware (10)
District of Columbia (11)
Florida (12)
Georgia (13)
Maryland (24)
North Carolina (37)
South Carolina (45)
Virginia (51)
West Virginia (54)

Division 6: East South Central

Alabama (01)
Kentucky (21)
Mississippi (28)
Tennessee (47)

Division 7: West South Central

Arkansas (05)
Louisiana (22)
Oklahoma (40)
Texas (48)

Region 4: West

Division 8: Mountain

Arizona (04)
Colorado (08)
Idaho (16)
New Mexico (35)
Montana (30)
Utah (49)
Nevada (32)
Wyoming (56)

Division 9: Pacific

Alaska (02)
California (06)
Hawaii (15)
Oregon (41)
Washington (53)

**Prior to June 1984, the Midwest Region was designated as the North Central Region.*

Appendix O. Percent of Genetic and Genomic Content in Areas of Study

The Percent of Perceived Role of Genetic and Genomic Content in Typical Areas of Study in a Speech-Language Pathology Program										
Perceived "No Role" of Genetics and Genomics in Areas of Study	Percent	f/n	Perceived "Some Role" of Genetics and Genomics in Areas of Study	Percent	f/n	Perceived "Moderate Role" of Genetics and Genomics in Areas of Study	Percent	f/n	Perceived "Significant Role" of Genetics and Genomics in Areas of Study	Percent	f/n	Perceived "Don't Know Role" of Genetics and Genomics in Areas of Study	Percent	f/n
Phonetic Acoustics	37%	19/51	Adult Language	5%	33/56	Reading	41	25/56	Hearing	61%	36/59	Speech Science	6%	3/54
Speech Science	24%	13/54	Swallowing	50%	29/58	Anatomy	41%	24/59	Autism	47%	28/59	Swallowing	5%	3/58
Adult Articulation	16%	9/56	Adult Articulation	48%	27/56	Pediatric Language	41%	24/59	Resonance	38%	21/55	Physical Assess	5%	3/60
Adult Language	11%	6/56	Feeding	46%	25/55	Pediatric Articulation	38%	23/60	Anatomy	37%	22/59	Attention	5%	3/57
Therapeutic Management	7%	4/59	Phonetic Acoustics	45%	23/51	Physical Assess	37 %	22/60	Physical Assessment	37 %	22/60	Therapy	5%	3/59
Voice	6%	3/55	Voice	44%	24/55	Resonance	35%	19/55	Cognition	36%	21/59	Phonetic Acoustics	4 %	2/51
Swallowing	5%	3/58	Therapeutic	42%	25/59	Therapy	36%	21/59	Fluency	36%	20/56	Adult Articulatio	4%	2/56
Feeding	4%	2/55	Pediatric Articulation	42%	25/60	Voice	33%	18/55	Learning	36%	21/59	Fluency	4%	2/56
Attention	4%	2/57	Attention	40%	23/57	Fluency	30%	17/56	Reading	30%	17/56	Feeding	4 %	2/55
Learning	3%	2/59	Learning	39%	23/59	Autism	30%	18/59	Pediatric Language	25%	15/59	Adult Language	4 %	2/56
Cognition	2%	1/59	Speech Science	37%	20/54	Attention	28%	16/57	Attention	23%	13/57	Cognition	3%	2/59
Resonance	2%	1/55	Cognition	36%	21/59	Swallowing	28%	16/58	Speech Science	22%	12/54	Voice	2%	1/55
Fluency	2%	1/56	Pediatric Language	31%	18/59	Feeding	27%	15/55	Feeding	20%	11/55	Reading	2%	1/56
Pediatric Language	2%	1/59	Fluency	29%	16/56	Adult Language	25%	14/56	Pediatric Articulation	18%	11/60	Pediatric Language	2%	1/59

Appendix P. Text Responses Provided by Speech-Language Pathologists

Question 2

	In your opinion, what impact does the current field of genetics/genomics have on speech-language pathology?
1.	"I am a fluency specialist. The current genetic research that is being done relevant to stuttering is shedding light on a very complex disorder."
2.	"Understanding autism, gene therapy for Parkinsons etc."
3.	"Knowing if there is a genetic disorder will affect treatment strategies and anticipated outcomes."
4.	"I understand that a gene for apraxia has been identified and studies of genetics can and has made contributions toward the conditions of cleft palate, autism, apraxia, and so forth."
5.	"new findings on genetic underpinnings of stuttering, autism have an effect on the approach to intervention."
6.	"Many current clients have had manifestations of a variety of speech/language and nasopharyngeal issues similar to a parent's childhood experiences."
7.	"Current impact is minimal, but the opportunity for a greater impact is hopefully on the horizon."
8.	"Needs further research. Much I've read is not definitive."
9.	"I think it should have more impact than I think it has. Families always ask "why" when a feeding or a communication disorder is identified. Genetic information may be useful in responding to their questions."
10.	"for example, knowing more about 22q11."
11.	"VCF was discovered by a speech pathologist who noted symptoms that children had in common-about 1978 or so."
12.	"I am currently working (and have always worked) in an adult setting, so this may bias my opinion on the matter. I feel that my opinion of may be different if I worked with children with developmental disorders rather than adults with acquired disorders. If this were the case, I may have rated genetics/genomics as having a greater impact on the field of SLP. As it is now, I believe that the field of genetics/genomics doesn't really influence what I do on a daily basis."
13.	"Already have babies surviving with syndromes, prematurities and associated disorders and disabilities. Genomics could effect increased survival, either repairing genes to alleviate syndromes or perhaps better survival, but a different or increased array of speech/communication issues...."
14.	Especially FOX2
15.	Not relevant to my practice.
16.	While I am unaware of specific advances in this area, I do believe that genetics plays a huge roll in various syndromes/disabilities with which many of the children enrolled in speech therapy programs are currently afflicted with.
17.	The research relating to Autism Spectrum Disorders and ADHD has helped drive and refine my intervention practices.
18.	Not sure how to answer this - depends on what aspects of speech-language pathology you refer to - clinical practice? understanding in etiology? counseling? etc.
19.	What I've been exposed to indicates it may help with diagnosis, but not sure if treatments are impacted.

20	This is not new idea. I was involved in genetic studies in the 1960's
.	

21.	It's difficult in my field to see direct impacts. I work in a SNF setting, and most of the genetic influence I see relates to dementia, which is thought to have a genetic component, and results in a communicative/cognitive deficit as a result.
22.	I specialize in Asperger Disorder and Autism, and there are studies, which point to genetics combined with possible environmental triggers for some forms of ASD.
23.	It has been extremely helpful to be able to tell my stuttering patients that there is a genetic link for stuttering.
24.	Unless there are something can be done, or it may just provide some individuals' (who stutter) sense of relief.
25.	My focus in mainly ASD, I also have a child with ASD so I have followed a lot of the research regarding this disorder and the genetic connections. I would place ADHD/ADD in the same area of thought. Many of the families I work with ASD have a parent and or another child within the spectrum. I know there is a genetic as well as an environmental connection. Stuttering too is an area of interest and connections are being found there as well.
26.	It helps to guide our diagnostic and intervention plans with patients and assists us in obtaining referrals to other professionals as well as counsel families.
27.	Try to stay current on journals and online articles -- and NT Ties Science section.
28.	especially in the area of stuttering disorders- helps to understand the nature of the disorder
29.	I work with people with Alzheimer's, Parkinson's, Multiple Sclerosis etc, so I hope that modifying a genetic response may help minimize some or all the symptoms of these diseases.
30.	particularly in stuttering!
31.	At least it should have a significant impact, though all research in genetics/genomics should be carefully reviewed and understood before impacting therapies.
32.	Identifying specific traits as they relate to common symptoms determined for disordered positively diagnosed with genetic/genomic diagnostic methods should more easily enable us to diagnose the etiology of conditions, advance our knowledge of what to look for within those diagnosed conditions and eventually support easier reimbursement for treatment of diagnosed conditions with related speech and language symptoms through insurance, etc.
33.	There are so many other things to know about- genetics is at the bottom of my list.
34.	Research information related to genomics and communication disorders takes time to be translated to the field in any practical way.
35.	have not read about the specific positive impacts on S-L skills
36.	Assessment of children with genetic syndromes can provide information on best treatment practice, as well as best surgeries, if necessary.
38.	Research could allow us to know more about SLI and stuttering.
39.	But I'm guessing that "significant" is most likely correct.
40.	When so much of what we do is therapy/intervention based... I think genetic implications are most useful in the sense of understanding where manifestations come from more than how to treat it.

with regard to CVA, cerebral aneurysm, cleft palate, deafness, etc.

Question 7.

	Do you feel confident in your understanding of the current field of genetics or genomics as it applies to the filed of speech-language pathology?
1.	I read the news and scientific literature and follow up with anything relevant by pulling the source and reading the article. But this is something I enjoy doing. I might not even use what I learn, but I read when it relates to my field even it relates somewhat such as genetic discoveries in psychological and developmental disorders.
2.	I completed that undergraduate degree in 1978 - a lot has changed within this field. While I have the basic understandings, I need to be updated via continuing education.
3.	More related to my basis science background, not my SLP background.
4.	Specifically in the area of Autism Spectrum Disorders
5.	Again, this depends on what aspects of SLP you mean.
6.	My youngest child has Trisomy 21, which built on my base knowledge of genetics and then I was diagnosed with invasive ductal carcinoma and learned even more about genetics.
7.	Basic enough understanding to explain to a parent the importance of genetic counseling.
8.	The field of genetics and genomics moves fast and rapidly adds new information to the body of knowledge. From my college coursework, I feel I have a broad understanding of the basics, but the most I get on current findings are blurbs and nuggets here and there that I stumble on adventitiously.
9.	I have always been interested in genetics & when I had free choice in topic selection of papers in school I often geared those papers toward genetics. I love twin studies!
10.	When the experiment is not overly complicated, I get it.
11.	Independent research (not Wikipedia) has helped, as have CP conferences.
12.	I am able to recognize signs/symptoms of possible genetic differences because of my work on a craniofacial team for 23 years. I know where to look for information.
13.	Because I have a PhD and work with transgenic models

Question 9.

	Have you received questions from parents/patients/clients regarding genetic/genomic principles of basic science such as ways in which conditions or traits are inherited, risks for recurrence of a condition or trait, etc.?
1.	Doesn't come up in the clinical population I work with Aphasia/TBI
2.	In regards to stuttering.
3.	I provide therapy for young children and their families. I am often the first person that tells them their child needs services, may have a significant delay, etc. Often parents will ask whether they should pursue genetic counseling (when referred by pediatrics), or what a definition means. Usually I explain the process on a very basic (family friendly) level.
4.	Mostly articulation and autism.
5.	During feeding and craniofacial clinics as well as during interventions in the NICU
6.	I work with elementary age students with autism, and I believe many families have discussed basic science about the diagnosis with their physicians and clinicians prior to this age.
7.	I had a consultation with parents whose child has an extremely rare disease and sometimes I see patients who exhibit minor malformation (familial ankyloglossia or class III dental malocclusion)

8. Especially related to stuttering and autism.

9.	"does this run in families? My mother-in-law says my husband had the same problem as a child"
10.	Only with parents of clients w/ Cl.Pal.
11.	Not in the public school setting.
12.	For children with CL/CP, we have a geneticist on our Craniofacial Team & most of the time, the questions are directed to and answered by her or a genetics counselor. However, for other voice issues (such as voice tremor or SD), I have been asked about heredity as a factor.
13.	Primarily related to genetics as a predictor of stroke.
14.	Work with large population of Hearing Impaired children.
15.	Parents who have noticed a pattern of heredity have raised these questions.

Question 10.

Describe the characteristics of the instructor teaching with course with genetic/genomic content? Check all that apply.	
1.	“Team taught by Audiologists and Nurse- both with interest and study in genetics.”
2.	“Guest lectures”
3.	“with much craniofacial experience and with courses in genetics
4.	“instructor completed coursework in genetic counseling as part of doctoral degree’

Appendix Q. Glossary

Angelman Syndrome: Angelman syndrome is a genetic disorder with characteristic features that include severe speech impairment, developmental delay, intellectual disability, and ataxia (problems with movement and balance).

Autosomal dominant: A gene on one of the non-sex chromosomes that is always expressed, even if only one copy is present. The chance of passing the gene to offspring is 50% for each pregnancy.

Behavioral genetics: The study of genes that may influence behavior.

Candidate gene: A gene located in a chromosome region suspected of being involved in a disease. *See also:* protein

Chromosomes: Where genes are found. Chromosomes are the structures in cells that "package" genes and ensure their safe transfer into new cells. A person has 46 chromosomes, half of which were inherited from each parent.

CNTNAP2: This is a gene (a protein) that encodes a member of the neurexin family which functions in the nervous system as cell adhesion molecules and receptors. This gene has been associated with new cases of autism spectrum disorder.

Complex trait: Trait that has a genetic component that does not follow strict Mendelian inheritance. May involve the interaction of two or more genes or gene-environment interactions. *See also:*Mendelian inheritance

Complex disorder: A disease that involves multiple genetic and environmental factors. Obesity, heart disease, and schizophrenia are examples of diseases that have multiple causes.

Congenital: Any trait present at birth, whether the result of a genetic or nongenetic factor.

Deletion: A loss of part of the DNA from a chromosome; can lead to a disease or abnormality. *See also*: chromosome, mutation

DNA (deoxyribonucleic acid): The molecule that encodes genetic information. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.

DNA sequence: The relative order of base pairs, whether in a DNA fragment, gene, chromosome, or an entire genome.

Dominant: An allele that is almost always expressed, even if only one copy is present. *See also*: gene, genome

Double helix: The twisted-ladder shape that two linear strands of DNA assume when complementary nucleotides on opposing strands bond together.

Dyspraxia: *See*: Oral and verbal apraxia

Enzyme: A protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds but not altering the direction or nature of the reaction.

Epigenetic: The study of "environmental," or non-genetic, factors inside cells that influence the ways genes produce proteins.

FOXP2: Is a gene (Forkhead-box P2). Mutations in this gene have been associated with severe speech and language disorder. It is a gene found in many mammals exhibiting some form of vocal ability such as whales, bats, songbirds, etc. It appears to be important for modulating plasticity of neural circuits.

GNPTAB: Is a gene carried by all higher animals. It has been implicated in some forms of stuttering disorder.

Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule). *See also*: gene expression

Gene expression: The process by which a gene's coded information is converted into the structures present and operating in the cell. Expressed genes include those that are transcribed into mRNA and then translated into protein and those that are transcribed into RNA but not translated into protein (e.g., transfer and ribosomal RNAs).

Gene mutation: Is a permanent inheritable change in the DNA sequence of a genome. They can have no effect, alter the product of a gene, or prevent the gene from functioning properly or completely.

Fluorescence in situ hybridization (FISH): A physical mapping approach that uses fluorescein tags to detect hybridization of probes with metaphase chromosomes and with the less-condensed somatic interphase chromatin.

Gene therapy: An experimental procedure aimed at replacing, manipulating, or supplementing nonfunctional or misfunctioning genes with healthy genes.
See also: gene, inherit

Genetic code: The sequence of nucleotides, coded in triplets (codons) along the mRNA, that determines the sequence of amino acids in protein synthesis. A gene's DNA sequence can be used to predict the mRNA sequence, and the genetic code can in turn be used to predict the amino acid sequence.

Genetic counseling: Provides patients and their families with education and information about genetic-related conditions and helps them make informed decisions.

Genetic discrimination: Prejudice against those who have or are likely to develop an inherited disorder.

Genome: Is the entirety of an organisms' hereditary information encoded either in DNA or in RNA.

Genetic engineering: Altering the genetic material of cells or organisms to enable them to make new substances or perform new functions.

Genome project: Research and technology-development effort aimed at mapping and sequencing the genome of human beings and certain model organisms.

Genomics: The study of genes and their function.

Genotype: The genetic constitution of an organism, as distinguished from its physical appearance (its phenotype).

Gene Expression: The "turning on" of a gene. Most human genes are active, or turn on, only in certain cells under certain conditions. Genes for eye color are active in eye cells but not in stomach cells. Similarly, some genes may lie dormant for years and then turn on and become malignant late in life.

Genetics: The study of genes and how they are inherited. Traditionally, genetic studies have focused on one gene at a time, while genomics is the study of large numbers of genes.

Genome: A collection of genes. The human genome is the collection of human genes, just as the dog genome is the collection of dog genes. All living things have genomes. Plants, animals and bacteria included.

Genomics: The study of large numbers of genes, or genomes. Genetics, by contrast, tends to focus on one gene at a time.

Genotype (*n.*): The particular form of a gene a person has.

Genotype (*v.*): To determine, though a DNA test, the particular form of a gene a person has. For instance, Alzheimer's researchers may genotype a patient's DNA to learn which form or forms of the *APOE* gene the person has.

Human Genome Project (HGP): Formerly titled Human Genome Initiative.

Lysosomal: Are cellular organelles that contain acid hydrolase enzymes to break down waste materials and cellular debris. They are found in animal cells. Lysosomes digest excess or worn-out organelles, food particles, and engulf viruses or bacteria

Mendelian Disorder (Also called *Single-Gene Disorder*): A disease caused by a single gene that is inherited in a straightforward manner from parent or parents to child. Huntington's disease and cystic fibrosis are examples. The term "Mendelian" refers to Gregor Mendel, an Austrian who did pioneering work on genes and traits in ordinary garden peas by showing that a single trait, such as color, can be determined by a single gene. Compared to "complex diseases," Mendelian disorders are relatively rare.

MicroRNAs: Small molecules found in plants and animals that may regulate the activity of genes.

Mutation: Any heritable change in DNA sequence. *See also:* polymorphism

Neural substrates: A set of brain structures that underlie a specific behavior or psychological state.

Online Mendelian Index of Man: Is an online compendium of human genes and known diseases with a genetic component.

Oral apraxia *See also:* verbal apraxia

Pedigree: A family tree diagram that shows how a particular genetic trait or disease has been inherited. *See also:* inherit

Penetrance: The probability of a gene or genetic trait being expressed. "Complete" penetrance means the gene or genes for a trait are expressed in all the population who have the genes. "Incomplete" penetrance means the genetic trait is expressed in only part of the population. The percent penetrance also may change with the age range of the population.

Phenotype: The physical characteristics of an organism or the presence of a disease that may or may not be genetic. *See also:* genotype

Polymorphism: Difference in DNA sequence among individuals that may underlie differences in health. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for genetic linkage analysis.
See also: mutation

Phenotype: A physical trait such as red hair, or behavior such as anxiety. A phenotype results from the “expression” of a gene or genes.

Prader-Willi syndrome (PWS): PWS is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity.

Protein: A molecule that carries out the business of cells. Enzymes and hormones are types of proteins. Most proteins have folds and bends, and their three-dimensional structures allow them to interact with other proteins, forming dynamic networks.

Recessive gene: A gene, which will be expressed only if there are 2 identical copies or, for a male, if one copy is present on the X chromosome.

RNA (Ribonucleic acid): A chemical found in the nucleus and cytoplasm of cells; it plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA. There are several classes of RNA molecules, including messenger RNA, transfer RNA, ribosomal RNA, and other small RNAs, each serving a different purpose.

RNA: A molecule involved in manufacturing proteins that may also regulate the activity of genes.

ROBO1: Is a gene (Roundabout homolog 1 protein) implicated in a communication disorder of a Finnish family with severe dyslexia. Study of the phonological memory component of the language acquisition system suggests that ROBO1 polymorphisms are associated with functioning in this system.

Sequence (*n.*): The sequence of genetic “letters” in a piece of DNA. A short DNA sequence might be: ACGTACGTACGT

Sequence (*v.*): To determine the sequence of genetic “letters” in a piece of DNA or an entire human genome.

Sex chromosome: The X or Y chromosome in human beings that determines the sex of an individual. Females have two X chromosomes in diploid cells; males have an X and a Y chromosome. The sex chromosomes comprise the 23rd chromosome pair in a karyotype.
See also: autosome

Sex-linked: Traits or diseases associated with the X or Y chromosome; generally seen in males.
See also: gene, mutation, sex chromosome

Single nucleotide polymorphism (SNP): DNA sequence variations that occur when a single nucleotide (A, T, C, or G) in the genome sequence is altered.
See also: mutation, polymorphism, single gene disorder

Single-gene disorder: Hereditary disorder caused by a mutant allele of a single gene (e.g., Duchenne muscular dystrophy, retinoblastoma, sickle cell disease).
See also: polygenic disorders

Single-Gene Disorder (Also called *Mendelian Disorder*): A disease caused by a single gene that is inherited in a straightforward manner from parent or parents to child. Huntington’s disease and cystic fibrosis are examples. The term "Mendelian" refers to Gregor Mendel, the Austrian who did pioneering work on genes and traits in ordinary garden peas by showing that a single trait, such as color, can be determined by a single gene. Compared to “complex diseases,” Mendelian disorders are relatively rare.

Syndrome: The group or recognizable pattern of symptoms or abnormalities that indicate a particular trait or disease.

Trait: A physical characteristic, such as red hair, that has a genetic component.

Verbal apraxia: A speech disorder caused by damage to specific areas of the cerebrum. It is characterized by the inability to execute learned movements of the structures of the mouth and throat to produce sounds.

X chromosome: One of the two sex chromosomes, X and Y. *See also:* Y chromosome, sex chromosome

Y chromosome: One of the two sex chromosomes, X and Y. *See also;* X chromosome, sex chromosome