RETT SYNDROME: A PLACE FOR ANGELS

Deborah Ann Cox

Submitted in partial fulfillment of the requirements of the Master of Education in Special Education degree at the University of Alaska Southeast

RECOMMENDED: Redacted for Privacy

Thomas Scott Duke, Ph.D., Committee Chair

Redacted for Privacy

Jill Burkert, Ph.D.

Redacted for Privacy

Jennifer Ward, M.L.I.Sc.

Redacted for Privacy

APPROVED:

Dean, School of Education: Larry Harris, Ph.D.

7-14-09

Date

For

Selena Grace Coleman

and

Her loving family

The Problem

Rett syndrome is a thief! It robs little girls of their projected life. It lulls their families into a false sense of security while their little girls develop normally for 6 to 18 months. Then it insidiously robs them of their skills and abilities until they are trapped in a body that won't respond. These little girls are called "silent angels" (Hunter, 2007).

Rett syndrome (RS) was originally identified in 1966 by the Austrian neurologist Andreas Rett, but his research and findings were written in an obscure form of the German language the medical world could not and did not translate. It wasn't until 1983, that Rett syndrome was re-identified and labeled as its own disorder (Hunter, 2007). The Rett Syndrome Research Foundation (2006) summarizes the condition best with:

Rett syndrome is a debilitating neurological disorder diagnosed almost exclusively in females. Children with Rett syndrome appear to develop normally until 6 to 18 months of age when they enter a period of regression, losing speech and motor skills. Most develop repetitive hand movements, irregular breathing patterns, seizures and extreme motor control problems. Rett syndrome leaves its victims profoundly disabled, requiring maximum assistance with every aspect of daily living. There is no cure. (Retrieved October 14, 2008 from http://www.rsrf.org/about_rett_syndrome/)

Research is ever going with regards to Rett syndrome. What is known as of now is that Rett syndrome is caused by a mutation of the gene MECP2. It is not passed down in families and it knows no ethnic boundaries. The majority of Rett girls live to adulthood (RSRF, 2006). The male child doesn't usually survive birth with Rett syndrome.

Author's Experiences and Beliefs

As the author, I would like to clarify my biases through a brief expose of my experiences and beliefs regarding this research document on Rett syndrome. It is important for me to acknowledge that my life's experiences as well as my education and professional experiences influence every action and judgment I make about the children in my care.

I am a fifty-three year old woman who chose a career in teaching five years ago and also fifty years ago. I have always wanted to be a Mommy and a teacher. I spent my young adult life performing many different jobs in order to raise my three children as a single parent. Obviously as a family we had to meet our needs by keeping with the basics. All medical, educational and financial information had to be obtained through basic research and immediate experience.

Three obvious life stories come to mind when I think of how my experience will influence my teaching of children with special needs. First, my daughter and I both have asthma. When she was a baby I had to take what I knew from my prior knowledge of asthma (my own) and make life saving decisions about my daughter's breathing. When she was around five I told her that she was the only one who could feel what her lungs and bronchial tubes were doing and she would have to work with me to make the decisions. Until of course she was old enough to make the decisions for herself. She is now a nurse who helps others make decisions. Second, my middle son was diagnosed with dyslexia. My first thoughts (and much of my thinking today) were, what does that mean. After extensive research and a lot of trial and error with teaching interventions I discovered that my son had a cross-dominance. He graduated from the University of Maine in four years with two degrees - history and political science - Magna Cum Lade. Third, my oldest son was active and impulsive. I'm sure today he would be medicated, but I chose a different route. My son needed excessive exercise and physical work to be calm enough

to do school work or read. When he was eight I gave him his first 8 pound maul to split firewood that we heated our house with. He has a family, owns a home and works at a job that requires physical labor and mental calculations. Each child and each situation was so different just as each resolution was intricate and dynamic but without the research and knowledge it could have been like throwing dust-bunnies in the wind trying to find a path for my children to excel.

I started college shortly after my last child started college. I got my Bachelor's of Liberal Arts in Social Science and my Master's of Arts in Teaching; I am working towards my Master's of Education in Special Education. I am receiving my education from the University of Alaska Southeast in Juneau. I have just started my fourth year of teaching special education in the elementary setting. My first year was spent in a Tlingit village on an island in Southeast Alaska where the foremost disability was fetal alcohol spectrum disorder (FASD) and the resources were very limited. I am now teaching in Juneau at Riverbend Elementary with special needs children in third, fourth and fifth grade including my angel with Rett syndrome.

It is my opinion-professionally, ethically and morally-that each child in special education should be afforded an education designed to fit their individual needs. Of course the public school system doesn't always allow for such luxuries. But as long as I teach I will research and discover everything about my children's disabilities and design their education accordingly. So much of the time teachers are put into such time constraints, caseload overloads and excessive bureaucratic paperwork that time does not allow this research to take place and they do the best they can with educated guesses and multifaceted interventions. But just one tiny clue, one tiny flick of an eye can change everything for a child. Don't get me wrong I'm not for reinventing the wheel with each child but how will I know what interventions to use unless I research and discover. Selena presented me with the perfect example of this need to know each child and their

disability intimately.

Angel:

Selena Grace is eleven with soft eyes that speak of her soul. Selena developed just as most 'normal' little girls until she was eighteen months old. At this point Selena stopped progressing at a normal rate and started to lose major milestones of development (walking, talking, eating). After extensive tests and tears it was discovered that Selena had Rett syndrome.

I have had Selena on my caseload for two years. She has just started fifth grade so I will have her one more year before she moves on to middle school. She is gorgeous. She has wavy brown hair and very expressive eyes. She has no voluntary control over her muscles- all her muscles. Any muscle control she attempts to work through her thoughts usually fails her. I have watched her stare at her hands while she is trying to play with a ball and her hands just won't do what she wants. Girls with RS brains' stop making neuro-pathways so they must use the very few that they developed before they started their decline. Sometimes Selena can locate the pathways but for the most part she can't. Her body is spastic and out of her control.

Communication is another huge issue for my Selena. Without the use of her muscles she cannot talk. Her only means of communication is eye gaze. All her thoughts and everybody she loves secrets are literally trapped inside her non-working body.

Selena's immune system is compromised. She catches every virus that comes through school. Her lungs and bronchial tubes are a constant concern.

Selena has frequent seizures, enough that her doctor has inserted a vegal- stimulator. When she has a seizure we swipe a magnet over her neck and this activates her stimulator and will usually stop the seizure. When this doesn't work we put Valium into her stomach tube and she goes to sleep.

Selena doesn't have the muscle control to chew or swallow so she is feed through a tube directly into her stomach. She does however have a little use of her mouth so we love to give her appropriate snacks. Her favorite is pulverized pears, yum.

As Selena has aged so has the curvature of her spine. She has severe scoliosis. This curvature impedes Selena's ability to breathe and properly utilize her internal organs. Selena had surgery six weeks ago to attach two metal rods to her spine in an attempt to straighten the curves. Her recovery is long and arduous.

Loving Selena Grace is easy, caring for her a privilege but teaching her is my challenge. Her physical needs are met through my experience as an Emergency Medical Technician, her emotional needs are met through my experience as a mother and grandmother, and this research document will contribute to her educational/communicational needs.

The Purpose for my Research

My purpose for writing this research document is to give teachers of girls with Rett syndrome useful information on teaching girls with this uncommon disorder. As teachers we are trained and educated on many of the more common (or publicized) disorders: attention deficit hyperactivity disorder (ADHD), dyslexia, autism and Downs syndrome and FASD, to name a few. While information on these disorders is easily accessed in schools, libraries and on the web less common disorders like Rett syndrome are harder to find. It is easy enough to locate a definition for Rett Syndrome, but useful information about educational interventions and research based practices is much harder to access.

Research is a key component for preparing me as a special needs teacher to create an insightful and appropriate education for the children on my caseload, including Selena. Her daily care and education is a balancing act of physical, medical and educational needs that are

individual to only her. What I learned in college is just the tip of the iceberg when it comes to teaching children like Selena. The true application of Selena's physical, medical and educational needs is research like this which is necessary for each and every child and their disability. I could settle for no less.

In extending my research to the educational practices of girls with Rett syndrome, it is my foremost hope that this research will benefit other teachers in building individual education plans for their children, just as it has benefited me in teaching Selena.

Methods

Selection Criteria

The 52 articles and 1 book used in this research encompass the educational practices (research-based interventions) of professionals with Rett syndrome girls. I also chose a varied assortment of medical articles to further my understanding in the disorder. Articles relating to the psychological affects of Rett syndrome on the family were rejected to focus on education. These articles were found in professional journals relating to Rett syndrome between 1983 and 2008.

Search Procedures

The basic definition of the disorder Rett syndrome was located in *The Rett Syndrome Handbook* (Hunter, 2007) and on the web at the Rett Syndrome Research Foundation (www.rettsyndrome.org).

Database Searches

For the article research I conducted a Boolean search in four databases: (a) Educational Research Information Center (ERIC) (Ebscohost); (b) Professional Development Collection (Ebscohost); (c) Education Journals (ProQuest); and (d) Education Abstracts (OCLC). I used "Rett syndrome" as my keyword to search each database. I limited my searches to the education

(and limited medical aspects) of girls with Rett syndrome.

ERIC (Ebscohost)

A Boolean search was done using keywords "rett syndrome" and just looking at peer reviewed articles. The search pulled up 16 articles, only seven of which fit the criteria (Baptista, Mercadante, Macedo & Schwartzman, 2006; Kerr, Archer, Evans, Prescott & Gibbon, 2006; Koppenhaver, Erickson & Skotko, 2001; Lotan, Isakov, & Merrick, 2004; Meador, Derby, McLaughlin, Barretto & Weber, 2007; Wales, Charman & Mount, 2004; Woodyatt, Marinac, Darnell, Sigafoos & Halle, 2004). There were many articles that were geared toward the families and caretakers of young ladies with Rett syndrome and a broader selection of information regarding autism. While Rett syndrome falls into the autistic spectrum disorder it is very individual in it's education recommendations.

Professional Development Collection (Ebscohost)

A Boolean search was done here as well using the keywords "rett syndrome." The search only included "scholarly (peer reviewed) articles" which included many articles that the ERIC search presented before I used just a 'peer reviewed' search. Seventy-seven articles came up, 11 of which fit the criteria (Browder, Ahlgrim-Delzell, Courtade, Gibbs, & Flowers, 2008; Crow, 2008; Evans & Meyer, 1999; Gura, 1999; Hetzroni, Rubin & Konkol, 2002; Izbicki, 2007; Katsiyannis, Ellenburg, Acton, & Torrey, 2001; McDougall, Kerr & Espie, 2005; Skotko, Koppenhaver & Erickson, 2004; Woodyatt, Marinac, Darnell, Sigafoos & Halle, 2004; Petry & Maes, 2006). This search presented many related articles to different symptoms of the disorder (profound learning disabilities, sleep disorders, socially challenging behaviors), a lot of broad autism articles and articles pertaining to state assessments and employment. One was a duplicate article. The authors of the eleven articles are as follows:

Educational Journals (ProQuest)

With an advanced search in the database using the keywords "rett syndrome" there were 153 hits. These were all considered "scholarly journals." There seemed to be many articles on autism without the specifics of Rett syndrome. I narrowed the search to articles with only Rett syndrome in the title and that fit the criteria of which I found 27 (Bargerhuff, 2003; Bisland, 2003; Brehm, 2003; Colvin, et al., 2003; Dahle, 2003; Ellaway, et al., 1999; Ellaway, Buchholz, Smith, Leonard & Christodoulou, 1998; Ellaway, Sholler, Leonard & Christodoulou, 1999; Fiumara, Barone, D'Asaro, Manzullo & Pavone, 1998; Fyfe, Leonard, Dye & Leonard, 1999; Julu, et al., 2001; Kerr & Julu, 1999; Kohl, Schlosser & Sancibrian, 2001; Kozinetz, Skender, MacNaughton, del Junco & Yamamura, 1996; Pan, Lane, Heatherington, & Percy, 1999; Percy, 1999; Pizzamiglio et al., 2008; Price & Chang, 2000; Schanen, 1999; Schanen, et al., 1998; Schultz, Glaze, Motil, Hebert & Percy, 1997; Shetty, Chatters, Tilton & Lacassie 2000; Shukla, Kennedy & Cushing, 1999; Stallard, Williams, Lenton & Vellman, 2001; Umansky & Watson, 1997; Vanhala, Korhonen, Mikelsaar, Lindholm & Riikonen, 1998; Woodyatt, Sigafoos & Keen, 2002). There were many articles that didn't fit my criteria: vaccinations, fragile X, Asperger's, assorted autism and pervasive developmental disorder (PDD), Joubert syndrome, hearing loss, criminal justice, cerebral palsy and duplicates of articles I already have. This search provided me with the medical articles.

Education Abstracts (OCLC)

I used an advanced search with no limits using the keywords "rett syndrome" and hit on 47 articles of which I chose seven (Fyfe, et al.,2007; Mazzocco, et al., 1998; Mount, Charman, Hastings, Reilly & Cass, 2003; Novitsky, 2008; Talebizadeh, Bittel, Veatch, Kibiryeva & Butler, 2005; Zapella, Gillberg & Ehlers, 1998; Rapp, 2005;).

Coding Procedures

I have developed a coding procedure to categorize the information I have compiled in the 52 articles, books, annotated bibliographies and book reviews for this thesis. The coding form is based on: (a) disciplinary perspectives of author; (b) intended audience; (c) publication type; (d) research design; (e) participants; (f) data sources; (g) findings; and (e) emergent themes.

Disciplinary Perspective of Authors and Intended Audience

I felt it was important to the integrity of this research thesis to identify the disciplinary perspective of the authors as well as their intended audience. As a special needs teacher, am faced with gathering information from varied sources to compile into an education plan. While I limited my research to educators and medical professionals whose intended audiences were just their peers, I also wanted to utilize the familial perspective (on education) for these girls as their information was the catalyst to the educational and medical perspectives. I wanted to know how much information on the education of the girls was accessible through each venue: medical, education and familial.

Publication Type

I evaluated and identified each article by publication type (e.g. empirical study, descriptive article, position paper, guide, annotated bibliography). Empirical studies specifically state the author's methods for gathering and analyzing the qualitative and quantitative data. Descriptive articles describe experiences but do not specify methods for gathering and analyzing data. Position paper takes an explicit stand and advocates for a position. A guide will supply directions and/or strategies for implementation about the article's subject. An annotated bibliography is a list of books, articles or web sites that will supply information in an abstract form with a citation.

Research Design, Participants, Data Source, and Findings

I classified each empirical study according to its research design (i.e. quantitative research, qualitative research, mixed method research). I identified the subjects that participated in each study (e.g., girls with Rett syndrome, children with multiple disabilities, children with autism, children with PDD). I described the data source or sources each article used to gather data (i.e. surveys, designated length in time study, interviews, questionnaires). I then summarized the findings of each study.

Data Analysis/ Emergent Themes

Brown and Duke (2005) and Duke and McCarthy (2007) used the Stevick-Collaizi-Keen method of data analysis, highly reductive method of qualitative research inquiry, to extract and compile concepts, issues and themes for their respective qualitative studies. I, too, used that method for my research analysis.

Results

Disciplinary Perspectives, Intended Audience, and Publication Type

The authors, disciplinary perspective of the authors, intended audience and publication type are delineated in Table 1.

Table 1

Author(s)	Disciplinary Perspective of Author(s)	Intended Audience	Publication Type
Baptista, Mercadante, Macedo & Schwartzman, 2006	Medicine	All	Empirical Study
Bargerhuff, 2003	Education	Educators	Book Review
Bisland, 2003	Education	Educators	Book Review
Brehm, 2003	Education	Educators & School	Book Review

Author(s)	Disciplinary Perspective of Author(s)	Intended Audience	Publication Type
		Psychologists	
Browder, Ahlgrim-Delzell, Courtade, Gibbs & Flowers, 2008	Education	Educators	Empirical Study
Colvin, Fyfe, Leonard, Schiavello, Ellaway, De Klerk, Christodoulou, Msall, & Leonard, 2003	Medicine	Medical Professionals	Empirical Study
Crow, 2008	Education	All	Descriptive Article
Dahle, 2003	Education	Medical Professionals & Educators	Descriptive Article
Ellaway, Buchholz, Smith, Leonard & Christodoulou, 1998	Medicine	Medical Professionals	Empirical Study
Ellaway, Sholler, Leonard & Christodoulou, 1999	Medicine	Medical Professionals	Empirical Study
Ellaway, Williams, Leonard, Higgins, Wilcken & Christodoulou, 1999	Medicine	Medical Professionals	Empirical Study
Evans & Meyer, 1999	Education	Educators	Empirical Study
Fiumara, Barone, D'Asaro, Manzullo & Pavone, 1998	Medicine	Medical Professionals	Descriptive Article
Fyfe, Downs, McIlroy, Burford, Lister, Reilly, Laurvick, Philippe, Msall, Kaufmann, Ellaway & Leonard, 2007	Medicine	Medical Professionals & Educators	Empirical Study
Fyfe, Leonard, Dye & Leonard, 1999	Medicine	Medical Professionals	Empirical Study
Gura, 1999	Medicine	All	Descriptive

Author(s)	Disciplinary Perspective of Author(s)	Intended Audience	Publication Type
			Article
Hetzroni, Rubin & Konkol, 2002	Education	Educators & Care Providers	Empirical Study
Hunter, 2007	Parent	All	Book (Descriptive)
Izbicki, 2007	Medicine	All	Descriptive Article
Julu, Kerr, Apartopoulos, Al-Rawas, Witt-Engerstrom, Engerstrom, Jamal & Hansen, 2001	Medicine	Medical Professionals	Empirical Study
Katsiyannis, Ellenburg, Acton, & Torrey, 2001	Education	Educators	Guide
Kerr, Archer, Evans, Prescott & Gibbon, 2006	Medicine	Medical Professionals	Empirical Study
Kerr & Julu, 1999	Medicine	Medical Professionals	Descriptive Article
Kohl, Schlosser & Sancibrian, 2001	Education	Medical Professionals & Educators	Empirical Study
Koppenhaver, Erickson & Skotko, 2001	Education	Educators & Parents	Empirical Study
Kozinetz, Skender, MacNaughton, del Junco & Yamamura, 1996	Medicine	Medical Professionals	Empirical Study
Lotan, Isakov, & Merrick, 2004	Medicine	Medical Professionals	Empirical Study
Mazzocco, Pulsifer, Fiumara, Cocuzza, Nigro, Incorpora &	Medicine	Medical Professionals	Empirical Study

Author(s)	Disciplinary Perspective of Author(s)	Intended Audience	Publication Type
Barone, 1998			
McDougall, Kerr & Espie, 2005	Medicine	Medical Professionals	Empirical Study
Meador, Derby, McLaughlin, Barretto & Weber, 2007	Education	Educators & Care Providers	Empirical Study
Mount, Charman, Hastings, Reilly & Cass, 2003	Medicine	Medical Professionals	Empirical Study
Novitsky, 2008	Medicine	Medical Professionals, Educators & Parents	Descriptive Article
Pan, Lane, Heatherington, & Percy, 1999	Medicine	Medical Professionals	Empirical Study
Percy, 1999	Medicine	Medical Professionals	Descriptive Article
Petry & Maes, 2006	Medicine	Medical & Educators	Empirical Study
Pizzamiglio, Nasti, Piccardi, Zotti, Vitturini, Spitoni, Nanni, Guariglia & Morelli, 2008	Medicine	Medical Professionals	Empirical Study
Price & Chang, 2000	Education	Educators	Annotated Bibliography
Rapp, 2005	Medicine	Medical Professionals	Descriptive Article
Schanen, 1999	Medicine	Medical Professionals	Descriptive Article
Schanen, Kurezynski, Brunelle, Woodcock, Dure & Percy, 1998	Medicine	Medical Professionals	Descriptive Article
Schultz, Glaze, Motil, Hebert	Medicine	Medical	Empirical Study

Author(s)	Disciplinary Perspective of Author(s)	Intended Audience	Publication Type
& Percy, 1997		Professionals	
Shetty, Chatters, Tilton & Lacassie 2000	Medicine	Medical Professionals	Descriptive Article
Shukla, Kennedy & Cushing, 1999	Education	Educators	Empirical Study
Skotko, Koppenhaver & Erickson, 2004	Education	Educators & Care Providers	Empirical Study
Stallard, Williams, Lenton & Vellman, 2001	Medicine	Medical Professionals	Empirical Study
Talebizadeh, Bittel, Veatch, Kibiryeva & Butler, 2005	Medicine	Medical Professionals	Empirical Study
Umansky & Watson, 1997	Medicine	Medical Professionals	Empirical Study
Vanhala, Korhonen, Mikelsaar, Lindholm & Riikonen, 1998	Medicine	Medical Professionals	Empirical Study
Wales, Charman & Mount, 2004	Medicine	Medical Professionals	Empirical Study
Woodyatt, Marinac, Darnell, Sigafoos & Halle, 2004	Education	Educators	Empirical Study
Woodyatt, Sigafoos & Keen, 2002	Education	Educators	Empirical Study
Zapella, Gillberg & Ehlers, 1998	Medicine	Medical Professionals	Empirical Study

Disciplinary Perspectives

Thirty-five of the 52 items (67%) included in this literature review were written by medical doctors, either as practitioners or as researchers. Sixteen of the items (31%) were written by educators, either teachers or teachers of teachers. One book (1%) was written by a mother of a

girl with Rett syndrome.

Intended Audience

Twenty-eight of the 52 items (54%) included in the literature review were intended for medical professionals. Ten items (19%) were intended for educators. Four items (7%) were intended for both medical professionals and educators. Three items (5%) were intended for educators and care-providers. One item (2%) was intended for parents and educators. For parents alone there were no items but intended for all people regardless of profession were 6 items (11%).

Publication Type

Thirty-four of the 52 items (65%) of the literature reviewed were empirical studies. Twelve items (23%) were descriptive articles. Three items (5%) were book reviews. One item each (6%) were a guide, annotated bibliography and a book.

Research Design, Participants, Data Source, and Findings

I located thirty-four empirical studies that met my selection criteria. The research design, participants, data source and findings of each study are delineated in Table 2. Of the 34 articles empirical studies reviewed, 18 (53%) are qualitative studies, six articles (18%) are quantitative studies and ten articles (29%) are a mixture of both qualitative and quantitative. Most of the participants are girls with Rett syndrome with the exception of family involvement, children with autism and multiple disabilities and comparison groups of peers and subjects with fragile-X syndrome. Much of the data was collected by observation, questionnaires, experimentation of interventions and medical history. There are a few medical articles that involve blood and cerebrospinal fluids, treadmill and vital signs in increments and MRIs.

Table 2

Author(s)	Research Design	Participants	Data Sources	Findings
Baptista, Mercadante, Macedo & Schwartzman, 2006	Qualitative	7 girls with Rett syndrome (RS) (U.S.)	Eye-gaze technology	The data suggests that with the 7 girls that the intentional answering of questions involving picture identification, similarities identification and identifying the same, were significantly more right than wrong in their answers.
Browder, Ahlgrim-Delzell, Courtade, Gibbs, & Flowers, 2008	Qualitative	23 students from a self-contained classroom setting (southeast U.S)	Academic and Cognitive testing	Significant gains were indicated on the Early Literacy Skills Builder (ELSB).
Colvin, Fyfe, Leonard, Schiavello, Ellaway, De Klerk, Christodoulou, Msall, & Leonard, 2003	Quantitative	152 families from the Australian Paediatric Surveillance Unit	History of RS cases and follow-up questionnaires	Four scale findings: Kerr Scale (suggested scale of independence by Kerr and colleagues) mean score 22.9- maximum score 37, WeeFIM (Functional Independence Measure for Children-by observation of parent report) mean 29- max. 126 for

Author(s)	Research Design	Participants	Data Sources	Findings
				independence. Percy Scale (personal communication, regression, growth changes, air swallowed) mean 24.9- max 47. Pineda Scale (personal communication, social interaction) mean 16.3- max 31. All children affected with RS are severely functionally dependent.
Ellaway, Buchholz, Smith, Leonard & Christodoulou, 1998	Mixed methods	31 patients with RS (Australia)	Cytogenetic analyses	Girls with RS have a normal chromosome 15 (15q11-13) whereas Angelman syndrome is deeply affected on chromosome 15.
Ellaway, Sholler, Leonard & Christodoulou, 1999	Mixed methods	34 patients from the Australian Rett Register	12 lead ECG/ 24 hour Holter monitor	Nine of the girls presented with prolonged corrected QT values (0.45 to 0.53 seconds) to the other RS girls who were within normal ranges (0.33 to 0.45 seconds). The Holter monitor showed no

Author(s)	Research Design	Participants	Data Sources	Findings
				tachyarrhythmia.
Ellaway, Williams, Leonard, Higgins, Wilcken & Christodoulou, 1999	Qualitative	31 subjects from Australia	8-week treatment	More subjects improved than worsened on L-Carnitine on the behavioral and social subscale of the RS Motor Behavior questionnaire. More subjects significantly improved on the Well-Being Index and the Hand Apraxia scale.
Evans & Meyer, 1999	Mixed methods	1 girl with RS (New Zealand)	3-year observation	Positive, non-demanding, social interactions were associated with the most significant decreases in excess behaviors such as hand movement and rocking.
Fyfe, Downs, McIlroy, Burford, Lister, Reilly, Laurvick, Philippe, Msall, Kaufmann, Ellaway & Leonard, 2007	Qualitative	97 families with RS (Australia)	Video-taped observations and parental checklist	This study was developing a video-based assessment tool to be used in the home by the family to document girls with RS. This paper provided evidence that video-taping and family

Author(s)	Research Design	Participants	Data Sources	Findings
				questionnaires are a valid and feasible form to collect data on complex behavioral and functional components of RS.
Fyfe, Leonard, Dye & Leonard, 1999	Quantitative	79.4% of the Australian juvenile RS population in April 1995	Questionnaires for parents	The data was collected from equal amounts of RS case families and control families for four generations of miscarriages, birth defects and post-neonatal childhood deaths. Miscarriages and birth defects did not show a significant difference between groups. However, stillbirths in RS case families were nearly double that of the control families and nearly 2/3 of the RS case stillbirths were boys.
Hetzroni, Rubin & Konkol, 2002	Qualitative	3 girls with RS	Assessments, experiments with aug-com devices and observation	Girls with RS are capable of matching spoken words to symbols when provided with meaningful

Author(s)	Research Design	Participants	Data Sources	Findings
				instruction.
Julu, Kerr, Apartopoulos, Al-Rawas, Witt-Engerstrom, Engerstrom, Jamal & Hansen, 2001	Mixed methods	56 girls with RS	Recordings of breathing	Breathing rates are normal in during sleep but irregular during waking hours. Forced and apneustic (breath holding, deep inspirations, rapid expirations) are common among 5-10 year olds (inadequate breathing peaks between 10-18 years) and Valsalva (forcibly exhaled) or normal breathing is common in girls over 18 years.
Kerr, Archer, Evans, Prescott & Gibbon, 2006	Qualitative	13 girls with RS (some with classic RS and equal amount with atypical RS) (U.K.)	Medical history and questionnaires	The girls with 'classic' RS differ significantly than the 'atypical' children in severity of their disorder. This is shown in feeding difficulty, health, epilepsy, head circumference and age at onset of regression.
Kohl, Schlosser & Sancibrian, 2001	Qualitative	2 children with autism	Case studies and literature reviews	Clinicians and teachers are often confused at deciding graphic

Author(s)	Research Design	Participants	Data Sources	Findings
				or manual symbols to enhance language acquisition. There is not enough empirical studies to fall back on for research based techniques.
Kozinetz, Skender, MacNaughton, del Junco & Yamamura, 1996	Qualitative	NA	Literature review from different regions	RS most likely occurs in utero or prior to conception (a defective gene).
Lotan, Isakov, & Merrick, 2004	Qualitative	4 girls with RS (Israel)	Treadmill use and vital signs	Low intensity, daily treadmill training program can improve heart rates.
Mazzocco, Pulsifer, Fiumara, Cocuzza, Nigro, Incorpora & Barone, 1998	Qualitative	14 males with fragile-X, 12 girls with RS and 25 males and females in a comparison group (U.S.)	Cognitive testing and interviews	The difference in autistic behaviors in fragile-X and RS gives potential implications for appropriateness of either disorder being classified as PDD.
McDougall, Kerr & Espie, 2005	Qualitative	9 RS families (U.K.)	Questionnaires and interviews	Identified three themes relating to beliefs, coping and emotions. 1. RS itself, health issues, seizures, external triggers, developmental changes, medication and

Author(s)	Research Design	Participants	Data Sources	Findings
				unknown. 2. Selective responding, checking, ignoring, listening out, physical care, medication control child's sleep, external aids, out of bed, sleeping with child and a balance of cares and needs with waking her. 3. Responsibility of parent, feelings of support, dealing with emotions and emotional response.
Meador, Derby, McLaughlin, Barretto & Weber, 2007	Qualitative	1 girl with RS	Data collection and observation	This is a study of hand stereotypy and a response to external reinforcer (stimuli). While the child chose the book which overtly distracted her from her automatic response most of the time the results only lasted 36 seconds, whereas the choice of a donut bite lasted 66 seconds. Theory: the

Author(s)	Research Design	Participants	Data Sources	Findings
				donut provided the oral stimulation that the child needed from the stereotypy hand movement (to mouth).
Mount, Charman, Hastings, Reilly & Cass, 2003	Mixed methods	15 girls with RS and 15 individuals with severe mental retardation (SMR)	Questionnaire	6 of the 15 RS individuals (40%) showed a high probability of autism when tested/ 1 of the 15 individuals with SMR showed signs of autism.
Pan, Lane, Heatherington, & Percy, 1999	Mixed methods	6 girls with RS and 4 siblings	Proton Spectroscopic Brain Imaging at 4.1 Tesla	The ratio of creatine and *N*-acetylaspar- tate in girls with RS is significantly elevated in the white matter of the brain as compared to siblings without RS and a small elevation in the ratio of glutamate to *N*-acetylaspartate in the gray matter.
Petry & Maes, 2006	Mixed methods	6 participants with multiple disabilities	Video-taped observation and questionnaires	It was concluded that people with profound, multiple disabilities express the pleasure and displeasure

Author(s)	Research Design	Participants	Data Sources	Findings
				through sound and facial expression.
Pizzamiglio, Nasti, Piccardi, Zotti, Vitturini, Spitoni, Nanni, Guariglia & Morelli, 2008	Qualitative	1 girl with RS (Italy)	Measurement of behaviors before and after intervention	Motor skills age before was 11 months; after was 14 months. In cognitive skills two areas showed marked improvement: means-ends relations and object relations in space. In language skills the girl went from 2 words and 2 sentences-comprehension-to 58 words and 9 sentences. Behavioral skills showed significant global improvement.
Schultz, Glaze, Motil, Hebert & Percy, 1997	Quantitative	28 girls with RS (Texas)	Physical anatomy measurements	The rate of deceleration of foot growth relative to height/age is significant. The rate of hand growth relative to height/age is similar.
Shukla, Kennedy & Cushing, 1999	Mixed methods	3 students with multiple disabilities	Data collections and observations	Results suggest that peer support in the general education classroom is preferable to

Author(s)	Research Design	Participants	Data Sources	Findings
				direct support from a para-educator for children with severe multiple disabilities (including RS).
Skotko, Koppenhaver & Erickson, 2004	Qualitative	4 girls with RS	Video-taped observation	Phase I - record the typical ways mothers and daughters interact during story time. Most of the mothers dropped the direct commands for using the device to more questions for prediction and inference. They also sought more confirmation from their daughter's answers. Phase II - hand splinting the resting hand. Hand splinting seemed to allow for greater use of the assistive technology device. Phase III - the impact of a variety of assistive technologies. By the end of the study all four

Author(s)	Research Design	Participants	Data Sources	Findings
				girls were using a single- message device with greater than chance correct responses. Phase IV - evaluate the effects of parent training. The girls and their mothers increased their relationship in a positive manner with their communication interactions.
Stallard, Williams, Lenton & Vellman, 2001	Qualitative	34 parents	Pain diaries	Of the 34 children, twenty-five (73.5%) had experienced on at least one day moderate to severe pain. Twenty-three children (67.6%) had severe pain. Four children (11.7%) experienced pain for more than 30 minutes for more than 5 days. None of the children were receiving pain management.
Talebizadeh, Bittel, Veatch, Kibiryeva &	Quantitative	77 girls with autism and control female	Blood tests	Skewed X inactivation is more common in girls with autism

Author(s)	Research Design	Participants	Data Sources	Findings
Butler, 2005		siblings (U.S.)		than in control group. A higher percentage of mother of the girls with autism were detected to having X chromosome inactivation.
Umansky & Watson, 1997	Mixed methods	9 girls with RS (California)	Hand use experiments with objects – video-taped observations	Visual attention to objects was not a predictor of stereotypes hand movement nor was it a substitute for reaching and grasping an object. It also did not have an opposite affect of suppressing the hand movement. *Incidental finding: shifts in eye gaze (inattention) was a predecessor of stereotypies hand movement.
Vanhala, Korhonen, Mikelsaar, Lindholm & Riikonen, 1998	Quantitative	12 patients with RS and 12 in a control group (epilepsy and/or mental retardation)	Cerebrospinal fluid samples	Nerve Growth Factor was measured. In the RS group the mean was 1.8 pg/mL which is significantly lower than the control group where the mean was 7.0 pg/mL.

Author(s)	Research Design	Participants	Data Sources	Findings
Wales, Charman & Mount, 2004	Qualitative	8 girls with RS	Timed observation	Repetitive hand movement is not influenced by outside reinforcement.
Woodyatt, Marinac, Darnell, Sigafoos & Halle, 2004	Qualitative	8 girls with RS (U.S.)	Observation in classroom and play settings	Behavior states of girls with RS can be changed and improved with various interventions.
Woodyatt, Sigafoos & Keen, 2002	Qualitative	8 children with Autism	Interviews and observations	Teachers are able to interpret some paralinguistic acts and behaviors as forms of communication.
Zapella, Gillberg & Ehlers, 1998	Quantitative	30 girls with RS (Sweden & Italy)	Consultations	Because of the many variant degrees of RS it was concluded that RS should be categorized as its own entity; Rett Complex.

Emergent Themes

Two broad themes emerged from the analysis of the 52 items included in this review of the literature. These emergent themes include: (a) characteristics of Rett syndrome and medical research; and (b) education and related services. These two theme clusters and their associated formulated meanings are delineated in Table 3 and Table 4.

Table 3

Characteristics of Rett Syndrome and Medical Research
• 1 in 8,500 births results in RS. As of August 2008 there were 3,800 confirmed cases in North America. There is an estimated 30,000 cases in China of which only 200 have been diagnosed. Global awareness is crucial.
• October 1999, Zoghbi, Uta Franke, Amir and collegues report the mutation of the *MECP2* gene in nearly a third of all RS cases studied. They found this by locating the region around the q28 segment of the X chromosome and then by following up with Eric Hoffman's belief that it was located in the Xq28. With this information and an extreme case of 'process of elimination' Zoghbi found the *MECP2* gene.
• Video-taping and family questionnaires have provided valuable data regarding behavioral and functional components of RS. This is done in the home (naturalistic setting) rather than in a clinical setting. It affords families in rural communities to participate in studies of RS. It also can further the medical research of RS.
• Studies have shown that using a distracting mechanism such as a donut bite lasts longer to curb hand movement stereotypy than a book because of the oral stimulation.
• Reinforcement (other than self-stimulation) does not seem to be a driving force in repetitive hand movement but studies support a view that they are maintained by neurochemical and/or neurobiological processes.
• It was concluded, in one study that people with profound, multiple disabilities express the pleasure and displeasure through sound and facial expression.
• Medical professionals use the Diagnostic and Statistical Manual of Mental Disorders, text-revised 4th Edition (DSM-IV-TR) to diagnose children with autism. School systems use the Individuals with Disabilities Education Act: Part 300, Assistance to the states for the education of children with disabilities (IDEA: Part 300). This can cause confusion to parents and advocates of children with disabilities.
• The International Rett Syndrome Association (IRSA), established in 1984, in 2002 it had appropriated $35 million dollars from Congress for research in the field of RS.
• The *MECP2* gene fails to regulate target genes involved in the development of girls with RS. RS adversely affects the development of the breathing rhythm, autonomic system and brain stem which can contribute to sudden death.
• Children with severe cognitive disabilities are more susceptible to pain. They seldom have the communication skills to express the pain and therefore many (most) of these children are not treated for pain. There needs to be a proper pain assessment and plan developed for disabled children with pain.
• Expansion on the epidemiology of RS depending on the severity scale selected. There are variations in the abilities of girls with RS (identified with the *MECP2* mutation) even through all affected children are severely functionally dependent.
• RS is caused by mutations in X-linked *MECP2* gene which encodes the methyl-CpG-binding protein (MeCP2), leading to inappropriate expression of genes that are supposed to be

Characteristics of Rett Syndrome and Medical Research

silenced.

- Proton spectroscopic brain imaging at 4.1 Tesla shows significant changes in RS subjects and the control group. The ratio of creatine and *N*-acetylaspartate in girls with RS is significantly elevated in the white matter of the brain as compared to siblings without RS and a small elevation in the ratio of glutamate to *N*-acetylaspartate in the gray matter.
- There are 14 abnormalities in girls with RS breathing rhythms. Hyperventilation is one of the 'energetic' breathing rhythms. It can reduce oxygen levels in the brain and can cause hypoxia and low expired carbon dioxide levels. The unstable breathing along with very low cardiac vagal tone (neonatal level) is suggestive of an immature brain stem. The girls are capable of generating normal rhythms but they are incapable of sustaining them.
- In RS the brain volume is smaller than normal; most affected are the frontal parts, caudate nucleus and the mid-brain. Nerve Growth Factor (NGF) affects the neurons in the basal forebrain. Girls with RS have extremely low levels of NGF.
- Angelman syndrome and Rett syndrome have many of the same characteristics. Methylation analysis shows that while Angelman syndrome's critical region is chromosome 15 (15q11-13) but RS is not. Therefore differentiating between the two in diagnosis.
- Using Sartwell's incubation period model it was discovered that RS most likely occurs in utero or prior to conception supporting the findings of a mutation of a gene.
- The study of inherited neurodegenerative disorders in children is in its infancy, specific treatment is lacking. It is important to proceed in diagnosis with caution as children with different clinical presentations could represent different mutations in the same gene.
- Unstable and changing breathing patterns and inadequate autonomic system and brain stem function in RS suggests brain immaturity. This is linked to early monoaminergic (nervous system) disorder which presents possible targets for the *MECP2* gene intervention in the future.
- A 12-year-old girl was discovered to have microcephaly, mental retardation and tracheoesophageal fistula (MMT) and all the features associated with RS too. Does this child have two distinct disorders or a 'new' disorder?
- A photographic case study shows that a young girl had no signs or symptoms of RS until she was 24 months old and was sick with a febrile illness. Her onset and progression was rapid (10 days). Is there a connection between the illness and a genetic predisposition?
- In a study of families of RS girls the question came up in the data collection about the connection between RS and sudden infant death syndrome (SIDS). There was a higher prevalence of SIDS in the RS case families than in the control families. RS and SIDS have a similar component: sympathovagal imbalance. Could the SIDS deaths in the RS case families have been RS?
- It is not clear whether prolonged corrected QT values in girls with RS is a part of RS or if it is manifested from the immature central nervous system. It is also not clear whether it is to blame for sudden death in these girls.

Characteristics of Rett Syndrome and Medical Research
• Girls with RS have a plasma carnitine deficiency; this affects the mitochondrial energy production. One study suggests giving these girls L-Carnitine could benefit their symptoms of RS.
• A study was preformed on the medical history of two boys (with neonatal encephalopathy) born into families with recurrent RS. It is highly likely these boys have the same or similar gene mutation as the girls.
• Shift in gaze or inattention is linked to motivate stereotypies hand movement. Four theories are presented, 1) the void of focus is filled with self-stimulatory stereotypies, 2) the eye movement is part of the larger stereotypy, 3) the perceptual consequences of eye movement not the eye movement itself facilitates hand stereotypies, 4) the eye movement/ hand movement represents the developmental level of the girls with RS; as in infants who think of their hands as objects before they develop the ability to use their hands to grasp objects (approximately 3 months old).
• RS girls have small feet as compared to their height/age. Two theories are presented, 1) autonomic system abnormalities cause the hands and feet to be cold; possible lack of nutrients supplied by the blood reaching extremities, 2) increase motor dysfunction may influence foot size.
• Adults are under-diagnosed with RS. Patients with RS remain reasonably stable after the age of 28 instead of further regression. The stereotypical hand movements subside into a clasping of hands at midline. Most adults diagnosed with RS have the mutated *MECP2* gene.
• Girls with autism show skewed X inactivation. This may lead to more scientific understanding of the genetics of autism and in turn RS.
• It is highly accepted that girls with RS are severely mentally retarded. Is this a fair and accurate label considering the limited physical and communication skills? One study showed these girls abilities to think on a deeper level when afforded the appropriate communication device.
• Boys are seldom found to have RS. It is theorized (and confirmed in mice) that because boys are born with one X chromosome and one Y chromosome that their bodies are unable to compensate for a mutation of this type on the X chromosome. They therefore are stillborn or die shortly after birth.
• Girls with RS suffer with sleep disturbances. Parents interact in three distinct categories: beliefs about the sleep problems, coping with the sleep problems and the emotional factors. Further study is needed to study the attributes of sleep disturbances and their relationship to responses.
• There has been a study, on mice, that shows a reversal of symptoms of RS after targeting a specific gene.
• Some children with RS are able to speak, after the regression, they make up a distinct group of children with a mutation on the *MECP2* gene (atypical) that still have RS. The scale of symptoms presented are extremely varied. It is therefore concluded that many children (and/or adults) are misdiagnosed. RS is frequently mistaken for autism. Studies present that

Characteristics of Rett Syndrome and Medical Research
girls with RS desire and enjoy people and conversing, that is a very clear difference. • In a comparison of Fragile-X and RS as to which had more symptoms in common with autism (both are classified in the spectrum) RS was the one. The argument was for establishing a separate disorder for Fragile-X. • In a study performed with 30 Swedish and Italian girls with RS it was discovered that the presentation of the disorder has so many variations that it should have its own diagnosis category, Rett Complex.

Table 4

Education and Related Services
• Inclusion for children with severe multiple disabilities, in an age-appropriate general education classroom, provides important social, cognitive and emotional growth. • Children with developmental disorders (RS) present much less variance in social behavior than the general public. • Peer support in the general education classroom is shown to improve social competence, improve and model appropriate age-related behavior and improve academics for severe multiply disabled children (including girls with RS) as well as their peer supporters. • While research- based studies have been preformed to encourage early reading skills for children, little has been explored on teaching reading skills to children with severe mental disabilities. A new program and assessment was built to accommodate children with severe disabilities. The study that followed demonstrated the positive gains achieved through this program: Early Literacy Skills Builders and the assessment is Early Literacy Skills Assessment. • Books are underused for communication support. A study finds that when offered books and proper communication devices that young girls with RS can communicate properly with deeper thinking responses than otherwise thought possible. Each girl will need to be analyzed to the most appropriate communication device. It is important to assume competency and the ability to learn! • Making online learning acceptable to children with severe disabilities requires the utilization of assistive technology and Universal Design. Assistive technology is any item or system that improves function of people with disabilities. Universal Design is a design is making computers and programs accessible to people with or without disabilities. • Girls with RS are capable of matching spoken words to symbols when provided with meaningful instruction. Symbols were then placed around the classroom to encourage spontaneous communication. While anecdotal notes were taken of positive observation of this communication further study in this is required. • Teachers of girls with RS need to be vigilant in researching RS and the new research to apply

Education and Related Services
this to the girls' IEPs. The teacher needs to individualize her findings with the specifics to each individual child. Interventions change almost daily and should be a constant source of research. • Constant and reliable data collection is necessary to find appropriate interventions for girls with RS. The differing of degree of severity is a huge component for these interventions. Positive, non-demanding, social interactions were associated with the most significant decreases in excess behaviors. • The term Potential Communicative Act (PCA) describes acts and behaviors that many children with autism use to communicate before they acquire language (if they do). Teachers' interpretation is validated as an important component to create opportunities to enhance communication development. • It is imperative for therapists not to focus on isolated deficits in children with autism but to look at the child's needs, interests and preferences to initiate interventions. • The American Music Therapy Association has two publications that explore the avenue of music as an inclusive therapy for people with disabilities. • A sensory-motor rehabilitative program was put in place for an RS girl in Italy with a great outcome. Using Piaget's developmental stages activities were introduced to the girl in sequence of the stages. Improvement in her motor skills, cognitive skills, language skills and behavioral skills were documented. • Some but not all characteristics of autism were discovered in girls with RS. This overlapping of disorders can enhance learning and scientific possibilities. One can shed light on the other and vice versa. • Eye-gaze is an important tool in communication with girls with RS. Studies show that these girls are consistently more right than wrong in their answers with intentional gaze. • A low intensity, daily training program with ambulatory girls with RS can significantly improve heart rates. This program can be taught to para-educators by a physical therapist to add to a daily program. With the advancement of stages there is an increase of longer corrected QT intervals and T-wave changes; this causes fear of aerobic exercise because its been connected to sudden death.

Discussion

In this section, I will discuss the two major themes that I emerged from my research of the 52 articles and books included in this review of the literature.

Theme 1: Characteristics of Rett Syndrome and Medical Research

One in 8,500 to 10,000 births results in Rett syndrome. As of August 2008 there were

3,800 confirmed cases in North America. This neurodevelopmental disorder is not tied to any ethnicity or socioeconomic population; it strikes little girls indiscriminately across the world . There is an estimated 30,000 cases in China of which only 200 have been diagnosed. This is a global issue which requires global attention. The International Rett Syndrome Association (IRSA), established in 1984, appropriated $35 million dollars from Congress for research in the field of RS in 2002.

Sartwell's incubation period model was used to discover that RS most likely occurs in utero or prior to conception supporting the idea that it is caused by a mutation of a gene. In October 1999, Zoghbi, Uta Franke, Amir and colleagues reported that they found that the mutation of the *MECP2* gene is apparent in nearly a third of all RS cases studied. They located the region around the q28 segment of the X chromosome and then they followed up with Eric Hoffman's belief that it was located in the Xq28. With this information and an extreme case of 'process of elimination' Zoghbi found the *MECP2* gene and its relative affect on girls with RS. The gene *MECP2* encodes the methyl-CpG-binding protein (MeCP2). The encoding regulates the development (starting, stopping and rate of growth) in the body. Therefore the mutation leads to inappropriate expression of genes that are supposed to be silenced and/or awakened (targeting genes responsible for development). This adversely affects the child's global development: the autonomic system (breathing rhythms), the brain (growth and systems), the body (skeletal, muscular and vessels) and the nervous system; any of which can cause sudden death if compromised.

Boys are seldom found to have RS. It is theorized (and confirmed in mice) that because boys are born with one X chromosome and one Y chromosome that their bodies are unable to compensate (like girls with two X chromosomes) for a mutation of this type on the X

chromosome. They therefore are stillborn or die shortly after birth. Stillbirths in RS case families was nearly double that of other families and nearly two-thirds of the RS case stillbirths were boys. One study was preformed on the medical history of two boys (with neonatal encephalopathy) who were born into families with recurrent RS. It is highly likely that these boys have the same or similar gene mutation as the girls with RS.

Head size in girls with RS begins to grow at a normal pace until stage one of RS when growth slows down. Brain volume is smaller than normal in girls with RS too; most affected are the frontal parts, caudate nucleus and the mid-brain. Girls with RS have extremely low levels of Nerve Growth Factor. Nerve Growth Factor affects the neurons in the basal forebrain. Unstable and changing breathing patterns in connection with inadequate autonomic system and brain stem function in RS suggests brain immaturity. Proton spectroscopic brain imaging at 4.1 Tesla shows significant differences in the white and gray matter of the brain in RS girls as compared to siblings without RS. The ratio of creatine and *N*-acetylaspartate in girls with RS is significantly elevated in the white matter of the brain and a small elevation in the ratio of glutamate to *N*-acetylaspartate in the gray matter. The size of the brain and the chemical anomalies lead to a highly accepted theory that girls with RS are severely mentally retarded.

Fourteen abnormalities in girls with RS breathing rhythms have been identified. Forced and apneustic (breath holding, deep inspirations, rapid expirations) are common among 5-10 year olds (inadequate breathing peaks between 10-18 years) and Valsalva (forcibly exhaled) or normal breathing is common in girls over 18 years. Hyperventilation, deep breathing, rapid shallow breathing and tachypnoea constitute 'energetic breathing.' It can reduce oxygen levels in the brain and can cause hypoxia and low expired carbon dioxide levels. The unstable breathing along with very low cardiac vagal tone (neonatal level) is suggestive of an immature brain stem.

Girls with RS are capable of generating normal rhythms but they are incapable of sustaining them. Breathing rates are normal in during sleep but irregular during waking hours. They also tend to level out to a more normal rhythm in adulthood.

Along with the immature brain stem there is evidence of an immature central nervous system. It is not clear whether prolonged corrected QT values in girls with RS are a part of RS or if it is manifested from that immaturity. It is also not clear whether it is to blame for sudden death in these girls. This is linked to early monoaminergic (nervous system) disorder which presents possible targets for the *MECP2* gene intervention in the future.

Stereotypical hand movements of wringing the hands and or putting the hands in the mouth are a diagnosing sign of RS. There have been some studies on the nature of these movements and what might intensify or lessen the act. The consensus from the articles and books in this study are that the hand movements are caused by neurochemical and/or neurobiological processes. Oral stimulation is also a strong theory and backed up by a study which provided food as a reinforcer. There seems to be more hand action when the child is stressed or excited and an incidental finding through the studies is that shifts in eye gaze (inattention) was a predecessor of stereotypies hand movement. One theory of interest is that the eye movement/ hand movement represents the developmental level of the girls with RS; as in infants who think of their hands as objects before they develop the ability to use their hands to grasp objects (approximately 3 months old). This theory follows Piaget's stages of development.

RS has many variations in the abilities of the girls even though most of the girls are severely functionally dependent. There are also variations of the onset of RS. A photographic case study shows that a young girl had no signs or symptoms of RS until she was 24 months old and was sick with a febrile illness. Her onset and progression was rapid (10 days). Some girls

with RS are able to speak, after the regression; they make up a distinct group of children with a mutation on the *MECP2* gene (atypical) that still have RS. The girls with 'classic' RS differ significantly than the 'atypical' children in severity of their disorder. This is shown in feeding difficulty, health, epilepsy, head circumference and age at onset of regression. The variations cause many children (and/or adults) to be misdiagnosed. Patients with RS remain reasonably stable after the age of 28 instead of having further regression. The stereotypical hand movements subside into a clasping of hands at midline. Most adults diagnosed with RS have the mutated *MECP2* gene.

Diagnosis at this stage is in the signs and symptoms quickly followed by the genetic testing. Before the genetic testing was an option RS was classified on the spectrum of autism. Medical professionals use the Diagnostic and Statistical Manual of Mental Disorders, text-revised 4th Edition (DSM-IV-TR) to diagnose children with autism. While there are many similarities with RS and autism one difference stands out, girls with RS desire and enjoy people and conversing. There are many disorders that are similar to RS (and in turn, autism) which causes a question of RS having its own individual disorder. Angelman syndrome and RS have many of the same characteristics. Methylation analysis shows that Angelman syndrome's critical region is chromosome 15 (15q11-13) but RS is not; therefore differentiating between the two in diagnosis. A 12 year old girl was discovered to have microcephaly, mental retardation and tracheoesophageal fistula (MMT) plus all the features associated with RS too. Does this child have two distinct disorders or a 'new' disorder? The question came up in the data collection of the connection between RS and sudden infant death syndrome (SIDS). There was a higher prevalence of SIDS in the RS case families than in the control families. RS and SIDS have a similar component: sympathovagal imbalance. Fragile-X and RS both have similar

characteristics to autism (both are on the spectrum) with RS have more. There is an argument for Fragile-X to establish its own disorder. In a study performed with thirty Swedish and Italian girls with RS it was discovered that the presentation of the disorder has so many variations that it should have its own diagnosis category; Rett Complex.

As a result of this research study I have a much clearer picture of Selena's disorder. She is globally affected and I now understand more thoroughly why she is acting and doing the things her little body does. Everything from her sleep disturbances, her seizures, her breathing issues, her communication, her muscle spasms, her eating, her mobility and so much more was spelled out for me as a care-provider for her. I have become more in-tuned to her needs, her wants and her possibilities.

Accurate data collection, anecdotal notes, pictures of multiple strategies used with Selena in her day-to-day school life could become a piece of the larger puzzle called Rett syndrome. Participation in conferences and in on-line discussions of my findings could benefit the growing scientific data and information highway that has started with recent medical interest in this disorder. Further (daily) research collection is necessary to stay informed and now I have the means and the know-how to do this research.

Theme 2: Education and Related Services for Students with Rett Syndrome

Girls with RS are capable of learning. They are to be afforded a Free and Appropriate Education (FAPE) just like every other child in America. While the medical community uses the DSM-IV-TR to diagnose children (including girls with RS) the education system uses the Individuals with Disabilities Education Act: Part 300, Assistance to the states for the education of children with disabilities (IDEA: Part 300). This gives teachers and parents a strong say in these girls' education. While inclusion and peer support for children with severe multiple

disabilities (RS), in an age-appropriate general education classroom, provides important social, cognitive and emotional growth, studies also reveal that we as educators and parents must always consider the least restrictive environment for each girl and their variations of this disorder, which at time includes one-on-one with a teacher (or related service provider) and inclusion. Each individual girl with RS will differ and vary in their strengths and weaknesses (as all children do) so it is imperative that the teacher be vigilant in their assessments (formative and summative) and flexible to change as interventions can change almost daily. This change also has much to do with the progress of research in the field of RS. Teachers must stay in constant touch with the RS community to keep her/his Individual Education Plan (IEP) up to date. The teacher must also become part of the community of RS with accurate and reliable data collection and professional insight into the world of girls with RS.

First and foremost girls with RS struggle to communicate. They are locked in bodies that don't function when the girls want them to. With the many variations of RS a girl might learn to speak but many rely on eye-gaze as their only form of communication. For many, eye-gaze is the only way to communicate their wants and needs and the only way to show signs of learning. Studies show that these girls are consistently more right than wrong in their answers with intentional gaze. The term Potential Communicative Act (PCA) describes acts and behaviors that many children with autism use to communicate before they acquire language (if they do). Teachers' interpretation is validated as an important component to create opportunities to enhance communication development. Girls with RS are capable of matching spoken words to symbols when provided with meaningful instruction.

Books are underused for communication support. One of the studies I investigated finds that when offered books and proper communication devices that young girls with RS can

communicate properly with deeper thinking responses than otherwise thought possible. Augmentative communication devices come in all shapes and forms, from low-tech to extremely high-tech. A device can be as simple as a yes and no card (long narrow card with 'yes' and a happy face on one end and 'no' and a frowning face on the other), a small white board with two to six choices written on it, a baking pan with symbolic pictures and words with magnets and the list can continue along with the imagination of the girls care-takers. Girls with RS are capable of matching spoken words to symbols when provided with meaningful instruction. Or a device can be as complex as a computerized device that responds (after appropriate input) to the girls eye-gaze or computer programs that respond to eye-gaze or movement. Making online learning acceptable to children with severe disabilities requires the utilization of assistive technology and Universal Design. Assistive technology is any item or system that improves function of people with disabilities. Universal Design is a design is making computers and programs accessible to people with or without disabilities.

When the government produced the No Child Left Behind Act (NCLB) schools actively participated in and preformed research- based studies to encourage early reading skills for children. Little was explored on teaching reading skills to children with moderate to severe mental disabilities and/or children with little or no communication skills. Each teacher was expected to accommodate research-based programs and curriculum to fit the needs of these special needs children. This is a Herculean task, especially for children with low or no communication skills. A group of teachers and researchers got together and produced a program and an assessment for this program for these particular children: Early Literacy Skills Builders and the Early Literacy Skills Assessment. As mentioned earlier, girls with RS can learn to respond to questions using deeper thinking answers when given the proper instruction and tools

to work with.

Another huge aspect of the educational day for girls with RS is motor function. Because of the many health and body issues that are part of RS it is important to keep these girls moving. One of the studies I read was about a sensory-motor rehabilitative program that was put in place for a girl with RS in Italy which had a great outcome. Using Piaget's developmental stages, activities were introduced to the girl in sequence of the stages. Improvement in her motor skills, cognitive skills, language skills and behavioral skills were documented. Another study showed that a low intensity, daily training program with ambulatory girls with RS can significantly improve heart rates. This program can be taught to para-educators by a physical therapist to add to a daily program.

I am anxious to try new research-based strategies with Selena. The program that was set up to further the usefulness of the girls' hands using Piaget's stages of development was fabulous. The peer support article will provide Selena with a support group as she transitions from elementary school to middle school and possibly on into high school then college or adult life. There are many research-based interventions here to try, use and document. Selena will also be receiving a new, state-of-the-art aug-com device which should open many doors of communication for her. Who knows perhaps in my experiences with Selena we may come up with new strategies that will work for other girls like her.

Conclusion

The study of RS is in its infancy but it is gathering momentum. In the articles I reviewed I would approximate that three-fourths of them were somewhat obsolete by the time I read them. While I was researching The Rett Syndrome Handbook they also came out with a new edition. From 1983 to today the amounts and types of studies have made significant changes in what we

know about RS. The discovery of the mutation of gene *MECP2* was probably the most significant recent change and it just got the party started. Each new discovery brings on more questions. Is it a fair and accurate to label girls with RS as severely mentally retarded considering the limited physical and communication skills and lack of appropriate assessments? Is there a connection between the onset of RS and illness? Should RS be its own disorder or should it stay on the autism spectrum? How much can we teach our girls with RS? When will they complete the study on the computer program where you can think a letter and your computer will type it? How can we stop or reverse the symptoms of RS? Researchers have shown that they can give mice RS, how long will it take them to reverse this process? How can we help the girls sleep? When will they discover a medication to stop the seizures? The questions are endless…

I originally tried to single-in on the education of girls with RS but it became apparent to me early on that the medical, educational and familial aspects of RS are so intertwined at this point in time that it was impossible to separate them without skewing the integrity of my work. It was also obvious to me in my research that the education field needs to step up their game in offering these amazing girls a life that they deserve. I am sure that with families, medical professionals and educators working together and collecting and sharing data that all aspects of the girls' lives will come together.

As for Selena Grace, she is the light of my day. While she has the 'typical' classic RS at the most severe range she has a smile for me each day. I have conversations with her that I know she understands. This understanding comes in the form of communication that is shared without words. More expressive eyes I have never seen. I can calm her and she certainly can calm me. I understand and accept her instinctual need to put her hands in her mouth and I give her time. I

understand and stand ready to serve her fragile health needs. I understand her need as well as her frustration with communication and I will be ever vigilant in finding the best device for her. I understand and accommodate her need (as an early adolescent) to be moody. I understand the importance to her to be around friends her own age. I understand that she expects me to teach her and I expect her to learn. But most of all I understand that Selena Grace was brought into my life for a reason and every day that I'm with her there is proof. I am a better person and teacher for knowing her.

References

Baptista, P., Mercadante, M., Macedo, E., & Schwartzman, J. (2006). Cognitive performance in Rett syndrome girls: A pilot study using eye tracking technology. *Journal of Intellectual Disability Research, 50*(9), 662-666.

Bargerhuff, M. E. (2003). A book review. *Remedial and Special Education, 24* (4), 255-256. Published regarding, Murray-Slutsky, C. & Paris, B. A. (2000). *Exploring the spectrum of autism and pervasive developmental disorders: Intervention strategies.* San Antonio, TX: Psychology Corp.

Bisland, A. (2003). A book review. *Remedial and Special Education, 24* (2), 127-128. Published regarding, Molfese, D. L. & Molfese, V. J. (Eds). (2002). *Developing variations in learning: Applications to social, executive function, language and reading skills.* Mahwah, NJ.: Erlbaum.

Brehm, K. (2003). Book Review Section. Lessons to be learned at the end of the day. *School Psychology Quarterly, 18* (1), 88-97. Published regarding, Grenot- Scheyer, M., Fisher, M., & Staub, D. (Eds.). (2001). *At the end of the day:Lessons learned in inclusive education.* Baltimore, MD: Paul H. Brookes Publishing Company.

Browder, D., Ahlgrim-Delzell, L., Courtade, O., Gibbs, S., & Flowers, C. (2008). Evaluation of the effectiveness of an early literacy program for students with significant developmental disabilities. *Exceptional Children, 75*(1), 33-52.

Brown, J. D., & Duke, T. S. (2005). Librarian and faculty collaborative instruction: A phenomenological self-study. *Research Strategies, 20*(3), 171-190.

Colvin, L., Fyfe, S., Leonard, S., Schiavello, T., Ellaway, C., De Klerk, N., Christodoulou, J., Msall, M. & Leonard, H., (2003). Describing the phenotype in Rett syndrome using a

population database. *Archives of Disease in Childhood, 88,* 38-43.

Crow, K. (2008). Four types of disabilities: Their impact on online learning. *TechTrends: Linking Research & Practice to Improve Learning, 52*(1), 51-55.

Dahle, K. B., (2003). The clinical and educational systems: Differences and Similarities. *Focus on Autism and Other Developmental Disabilities, 18* (4), 238-246, 256.

Ellaway, C., Buchholz, T., Smith, A., Leonard, H. & Christodoulou, J., (1998). Rett syndrome: Significant clinical overlap with Angelman syndrome but not with methylation status. *Journal of Child Neurology, 15* (5), 448-451.

Ellaway, C. J., Sholler, G., Leonard, H. & Christodoulou, J., (1999). Prolonged QT interval in Rett syndrome. *Archives of Disease in Childhood, 80,* 470-472.

Ellaway, C., Williams, K. Leonard, H., Higgins, G., Wilcken, B. & Christodoulou, J., (1999). Rett syndrome: Randomized controlled trial of L-Carnitine. *Journal of Child Neurology, 14* (3), 162-167.

Evans, I., & Meyer, L. (1999). Modifying adult interactional style as positive behavioural intervention for a child with Rett syndrome (n1). *Journal of Intellectual & Developmental Disability, 24*(3), 191.

Fiumara, A., Barone, R., D'Asero, G., Marzullo, E. & Pavone, L., (1998). Rett syndrome: Photographic evidence of rapid regression. *Journal of Child Neurology, 14* (8). 550-552.

Fyfe, S., Downs, J., McIlroy, O., Burford, B., Lister, J., Reilly, S., Laurvick, C. L., Philippe, C., Msall, M. Kaufmann, W. E., Ellaway, C. & Leonard, H. (2007). Development of a video-bases evaluation tool in Rett syndrome. *Journal of Autism and Developmental Disorders, 37* (9), 1636-1646.

Fyfe, S., Leonard, H., Dye, D. & Leonard, S., (1999). Patterns of pregnancy loss, perinatal mortality and post neonatal childhood deaths in families of girls with Rett syndrome. *Journal of Child Neurology, 14* (7). 440-444.

Gura, T. (1999). Gene defect linked to Rett syndrome. *Science, 286*(5437), 27.

Hetzroni, O., Rubin, C., & Konkol, O. (2002). The use of assistive technology for symbol identification by children with Rett syndrome. *Journal of Intellectual & Developmental Disability, 27*(1), 57-71.

Hunter, K. (2007). *The Rett syndrome handbook* (2nd ed.). Clinton, MD: International Rett Syndrome Foundation Publishing.

Izbicki, J. (2007). Higher ground. *Education Journal, 12* (6), 20.

Janesick, V. J. (2004). *"Stretching" exercises for qualitative researchers* (2nd ed.). Thousand Oaks, CA: Sage Publications, Inc.

Julu, P. O. O., Kerr, A. M., Apartopoulos, F. Al-Rawas, S., Witt- Engerstrom, I. Engerstrom, L., Jamal, G. A. & Hansen, S., (2001). Characterisation of breathing and associated central autonomic dysfunction in the Rett disorder. *Archives of Disease in Childhood, 85,* 29-37.

Katsiyannis, A., Ellenburg, J., Acton, O., & Torrey, G. (2001). Addressing the needs of students with Rett syndrome. *Teaching Exceptional Children, 33*(5), 74-78.

Kerr, A., Archer, H., Evans, J., Prescott, R., & Gibbon, F. (2006). People with MECP2 mutation-positive Rett disorder who converse. *Journal of Intellectual Disability Research, 50(*5), 386-394.

Kerr, A. M. & Julu, P. O. O., (1999). Recent insights into hyperventilation form the study of Rett syndrome. *Archives of Disease in Childhood, 80*, 384-387.

Kohl, R. K., Schlosser, R. W. & Sancibrian, S., (2001). Effects of symbol, referent and instructional variables on the acquisition of aided and unaided symbols by individuals with autism spectrum disorder. *Focus on Autism and Other Developmental Disabilities, 16* (3), 162-169.

Koppenhaver, D., Erickson, K., & Skotko, B. (2001). Supporting communication of girls with Rett syndrome and their mothers in storybook reading. *International Journal of Disability, Development & Education, 48*(4), 395-410.

Kozinetz, C. A., Skender, M. L., MacNaughton, N. L., del Junco, D. J. & Yamamura, Y., (1996). Rett syndrome: An analysis using Sartwell's incubation period model. *Journal of Child Neurology, 12* (6), 361-364.

Lotan, M., Isakov, E., & Merrick, J. (2004). Improving functional skills and physical fitness in children with Rett syndrome. *Journal of Intellectual Disability Research, 48*(8), 730-735.

Mazzocco, M. M., Pulsifer, M., Fiumara, A., Cocuzza, M. Nigro, F. Incorpora, G. & Barone, R. (1998). Brief report: Autistic behaviors among children with fragile X or Rett syndrome: Implications for the classification of pervasive developmental disorder. *Journal of Autism and Developmental Disorders, 28* (4), 321-328.

McCarthy, K. W., & Duke, T. S. (2007). The observation, documentation and shared reflection process: Preparing early childhood educators to teach in Alaska Native communities. *Journal of Early Childhood Teacher Education, 28*(2), 97-113.

McDougall, A., Kerr, A., & Espie, C. (2005). Sleep disturbance in children with Rett syndrome: A qualitative investigation of the parental experience. *Journal of Applied Research in Intellectual Disabilities, 18*(3), 201-215.

Meador, S., Derby, K., McLaughlin, T., Barretto, A., & Weber, K. (2007). Using response latency within a preference assessment. *Behavior Analyst Today, 8*(1), 63-69.

Mount, R. H., Charman, T., Hastings, R. P., Reilly, S. & Cass, H. (2003). Features of autism in Rett syndrome and severe mental retardation. *Journal of Autism and Developmental Disorders, 33* (4), 435-442.

Novitsky, S. (2008). What is Rett syndrome? *Exceptional Parent Magazine, 38* (8), 31- 32.

Pan, J. W., Lane, J. B., Heatherington, H. & Percy, A. K., (1999). Rett syndrome: 1H spectroscopic imaging at 4.1 teals. *Journal of Child Neurology, 14* (8), 524-528.

Percy, A. K., (1999). Inherited neurodegenerative disease: The evolution of our thinking. *Journal of Child Neurology, 14* (4), 256-261.

Petry, K., & Maes, B. (2006). Identifying expressions of pleasure and displeasure by persons with profound and multiple disabilities. *Journal of Intellectual & Developmental Disability, 31*(1), 28-38.

Pizzamiglio, M. R., Nasti, M., Piccardi, L., Zotti, A., Vitturini, C., Spitoni, G., Nanni, M. V., Guariglia, C. & Morelli, D., (2008). Sensory-motor rehabilitation in Rett syndrome. *Focus on Autism and Other Developmental Disabilities, 23* (1), 49-62.

Price, H. E. & Chang, E. C., (2000). An annotated bibliography of music education research journals. *Update,* 19-26.

Rapp, C. E. (Jr.) (2005). Rett Syndrome: A brief update and special features in adults. *Exceptional Parent Magazine, 35* (5), 69-72.

Rett Syndrome Research Foundation, (2006). Introduction to Rett syndrome. Retrieved October 14, 2008 from http://www.rsrf.org/about_rett_syndrome/

Schanen, N. C., (1999). Molecular approaches to the Rett syndrome gene. *Journal of Child Neurology, 14* (12), 806-814.

Schanen, N. C., Kurezynski, T. W. Brunelle, D., Woodcock, M. M., Dure, L.S. & Percy, A. K., (1998). Neonatal encephalopathy in two boys in families with recurrent Rett syndrome. *Journal of Child Neurology, 15* (3), 229-231.

Schultz, R., Glaze, D., Motil, K., Hebert, D. & Percy, A., (1997). Hand and foot growth failure in Rett syndrome. *Journal of Child Neurology, 13* (2), 71-74.

Shetty, A. K., Chatters, R., Tilton, A. H. & Lacassie, Y., (2000). Syndrome of microcephaly, mental retardation, and tracheoesophageal fistula associated with features of Rett syndrome. *Journal of Child Neurology, 13* (1), 61-63.

Shukla, S., Kennedy, C. H. & Cushing, L. S., (1999). Intermediate school students with severe disabilities: Supporting their social participation in general education classrooms. *Journal of Positive Behavior Interventions, 1* (3), 130-140.

Skotko, B., Koppenhaver, D., & Erickson, K. (2004). Parent reading behaviors and communication outcomes in girls with Rett syndrome. *Exceptional Children, 70*(2) 145-166.

Stallard, P., Williams, L., Lenton, S. & Vellman, R., (2001). Pain in cognitively impaired, non-communicating children. *Archives of Disease in Childhood, 85,* 460-462.

Talebizadeh, Z., Bittel, D. C., Veatch, O. J., Kibiryeva, N. & Butler, M. G. (2005). Brief report: Non-random X chromosome inactivation in females with autism. *Journal of Autism and Developmental Disorders, 35* (5), 675-681.

Umansky, R. & Watson, J. S., (1997). Influence of eye movements on Rett stereotypies: Evidence suggesting a stage-specific regression. *Journal of Child Neurology, 13* (4), 158-162.

Vanhala, R., Korhonen, L., Mikelsaar, M. Lindholm, D. & Riikonen, R., (1998). Neurotropic factors in cerebrospinal fluid and serum of patients with Rett syndrome. *Journal of Child Neurology, 10* (6), 399-404.

Wales, L., Charman, T., & Mount, R. (2004). An analogue assessment of repetitive hand behaviours in girls and young women with Rett syndrome. *Journal of Intellectual Disability Research, 48*(7), 672-678.

Woodyatt, G., Marinac, J., Darnell, R., Sigafoos, J., & Halle, J. (2004). Behaviour state analysis in Rett syndrome: Continuous data reliability measurement. *International Journal of Disability Development and Education, 51*(4), 383-400.

Woodyatt, G., Sigafoos, J. & Keen, D., (2002). Verifying teacher perceptions of the potential communicative acts of children with autism. *Communication Disorders Quarterly, 23,* (3), 133-142.

Zapella, M., Gillberg, C. & Ehlers, S. (1998). The preserved speech variant: A subgroup of the Rett complex: A clinical report of 30 cases. *Journal of Autism and Developmental Disorders, 28* (6), 519-526.