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ORIGINAL ARTICLE

Victor McKusick and his short course

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Abstract

The Short Course in Human and Mammalian Genetics and Genomics (aka the “Short Course” or the “Bar Harbor course”) is one of Victor McKusick's landmark contributions to medical genetics. Conceived in 1959 as a way to increase the contribution of genetic advances to medicine, it has directly affected more than 7000 students and 600 participating faculty from around the world. Now, more than 10 years after his death, it continues to be a vibrant disseminator of genetics, and genomics knowledge for medicine, a catalytic agent for ongoing research and a source of collegiality in our field. What an extraordinary gift!

KEYWORDS

education, Johns Hopkins, The Bar Harbor Course, The Jackson Laboratory

1 | INTRODUCTION

Sixty one years ago, human genetics was little more than a glimmer in the eye of medicine: the structure of DNA had been described by Watson and Crick only six years earlier (Watson & Crick, 1953); the correct number of human chromosomes (46) had been determined only three years earlier (Tjio & Levan, 1956); trisomy 21 as the cause of Down syndrome had been reported only a few months earlier (Lejeune et al., 1959); gene mapping methods were rudimentary; and the methods for DNA sequencing would not be published for another 17 years (Maxam & Gilbert, 1977; Sanger et al., 1977). Despite this embryonic state of human genetics, Victor McKusick recognized its potential for medicine. Two years earlier he had altered his medical career plans from cardiology to genetics and on 1 July 1957 with the blessings of the Chair of the Department of Medicine at Johns Hopkins, A. McGhee Harvey, M.D., he established the first medical genetics clinic in the United States, known locally as “the Moore Clinic” (McKusick, 2006). Shortly thereafter, in an effort to garner NIH support for this new enterprise, McKusick met Earl Green, Ph.D., then director of the Jackson Lab (JAX) and member of an NIH site visit team reviewing McKusick's nascent program at Hopkins. Noting his Maine origins and their common interests in mammalian genetics,

Green invited McKusick to visit the JAX in Bar Harbor, Maine. This invitation led to a fateful meeting over lunch at Testa's restaurant on North Main St in Bar Harbor in mid-July 1959 between Victor and John Fuller, a JAX staff scientist with an interest in behavioral genetics who also served as the JAX's director of education.

At this meeting, as documented in a subsequent letter (written 11 August 1959, see Appendix S1), McKusick laid out his plans for a “Short Course in Medical Genetics” to be held at the JAX where the “work in mammalian genetics” provided “an excellent base for human work.” The content of the Short Course (SC) would be aimed at medical school faculty to “aid their research and teaching” in medical genetics. The emphasis was very much on “teaching the teachers” so that the students would use what they learned at the SC to expand medical genetics teaching and thinking at their respective medical schools.

The plan materialized quickly; the first SC was held one year later (August 7–18, 1960) in Bar Harbor at Bowdoin College's Oakes Center located on the shores of Frenchman's Bay (Figure 1). Victor had obtained funding from the National Foundation March of Dimes as part of its program on birth defects (see Funding) (Fuller & McKusick, 1961; McKusick, 1972). The initial structure (four morning lectures, optional afternoon workshops and an evening lecture)

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FIGURE 1 Elizabeth (“Tibby”) Russell demonstrating a mouse at an early version of the SC at Atlantic Oakes

allowed time for presentation of considerable formal content as well as informal one-on-one interactions between students and faculty and has persisted to the present. In the first year, there were approximately 45 students and a faculty comprised of nine from Johns Hopkins (JH), 11 from JAX and eight “visiting” lecturers from various academic centers. In an invited critique of this first SC, the participant’s comments were encouraging (or as Victor said, the “favorable comments outweighed the negative”) and were used to make modifications in the subsequent 1961 version. And so, the sprout took root.

For the first six years, the content of the SC focused on medical genetics. Thereafter, from 1967–1977, the focus was on the mouse in the odd and on humans in the even years. In 1979, the 50th anniversary of JAX, the SC organizers (see Table 1) recognized an increasing convergence of medical and mouse genetics and from that year forward the course included a mix of human and mouse genetics, typically interwoven in each presentation. Over its’ history, the SC has been held in three venues: the Atlantic Oakes Conference Center in downtown Bar Harbor (1960–1962); the Emerson School, a local elementary school (1963–1971); and, then in its current location, the auditorium on the campus of the JAX, just south of Bar Harbor (1972–present). In 2005, the evening lectures were moved to a hotel conference room centrally located in downtown Bar Harbor.

Several excellent accounts of the SC have been published at various times of its history (Fuller & McKusick, 1961; Guethlein, 1990; McKusick, 1972; McKusick, 2006; McKusick et al., 1999) and in oral histories available at the JAX and Johns Hopkins (see The Jackson Laboratory Oral History Collection, 1986, UCLA Oral History Project, 2001). Here we briefly recount these details but, in the spirit of this compendium, we will add personal recollections and details to give the reader a more intimate account of the SC from the perspective of students, faculty and organizers. Throughout, we will mention the names of some of the prominent participants but our list is far from complete and we apologize to the many individuals who have played important roles in the SC over the years but whose names we were not able to include.

TABLE 1 Organizers of the short course over the years

Name	Institution	Years
Victor McKusick	Johns Hopkins	1960–2008
John Fuller	Jackson Lab	1960–1967
Elizabeth Russell	Jackson Lab	1967–1974
Seldon Bernstein	Jackson Lab	1968–1969
Thomas Roderick	Jackson Lab	1974–1990
Edward Birkenmeier	Jackson Lab	1990–1996
Jurgen Naggert	Jackson Lab	1998–2013
Patsy Nishina	Jackson Lab	1998–2013
David Valle	Johns Hopkins	1990–present
Aravinda Chakravarti	Johns Hopkins; NYU	2010–present
Charles Wray	Jackson Lab	2014–present
Charles Lee	Jackson Lab	2015–present
Ada Hamosh	Johns Hopkins	2017–present
Gregory Cox	Jackson Lab	2017–present
Nadia Rosenthal	Jackson Lab	2017–present

2 | GOALS AND SOME CHARACTERISTICS

From the outset, the primary goal of McKusick and Fuller was to educate at all levels a cohort of physician and investigator “students” who, in turn, would use this experience not only to inform their practice and their research but also to provide a fund of knowledge to enhance their own educational activities back at their respective schools—to “teach the teachers” was a frequently repeated mantra. In this way, McKusick planned to multiply the effect of the SC and ramp up knowledge and teaching of genetics in academic medical centers as quickly as possible.

From the very beginning, heterogeneity in the extent of training and the level of understanding of genetics has been a prominent characteristic of the students of the SC with pre-docs, post-docs, fellows, junior faculty and senior faculty all present in the same class. This feature has persisted and, as a consequence, effective lecturers start at a relatively basic level and gradually build complexity of content in their lectures, often culminating with the “latest and greatest” unpublished work from their lab or clinic. For many of us, this style of teaching has influenced our educational activities overall. Lecturing at the SC in the summer is good for pedagogy throughout the academic year.

Another key characteristic of the SC is the tradition that many of the faculty attend nearly all the lectures. This plus the heterogeneity of the students, means that there are often some in the auditorium who know as much or more about the topic than the lecturer. To the benefit of all involved, this keeps the lecturers on their toes, as evidenced by the considerable preparation and effort they put into their presentations. It also fosters lively discussions following the lectures.

A final point: when the venue moved to the auditorium at the JAX in 1972, the configuration of that relatively small auditorium with a capacity of 126 had subtle but clear effects on the experience of

lecturing at the SC. From the perspective of the lecturer looking out at the class, the podium has always been on the left and the space between the screen and the first row of students is compressed so that the lecturer is nearly in the audience with the rows of seats rising sharply to the back of the auditorium. Dr. McKusick was a continuous presence and always sat in the right-most seat on the first row, taking notes in his ever-present brown, pocket-sized notebooks. From that vantage point, he would frequently jump up to take pictures or ask questions. This location explains the awkward composition of the thousands of photographic images he made from that vantage point (see Figure 2). Following his death in 2008, McKusick's chair was labeled with a plaque recognizing the many years he used it as a platform to teach and to soak up his favorite topic. Out of habit, other longtime faculty members sat reproducibly in specific locations in the auditorium. John Phillips sits typically in the upper left corner asking questions after nearly every lecture. Laird Jackson sat in the upper right corner and gave words of encouragement to speakers as they completed their lectures and left the room through the door at the upper right. Aravinda Chakravarti sits in the middle of the back row and asks questions designed to keep speakers honest. Ken Paigen could be found midway up the right side of the auditorium, from where he asked many penetrating questions.

3 | STRUCTURE

As mentioned above, the basic structure of the Short Course has remained unchanged from its inception: the morning bell rings at 8:30 and is followed by four 50 min lectures (~40 min with 10 min of questions). A 30-min coffee break is positioned between the second and third lectures providing time for student and faculty interactions. The morning talks end at 12:30 and are followed by an onsite lunch with tables posted for each of the morning's speakers so that interested students can ask additional questions and otherwise interact with speakers over lunch. An optional afternoon workshop follows from 1:30 to 3:30. A single "evening lecture" or similar designated event takes place from 7:30 to 9. These presentations typically are given by leaders in the field. In recent years, three of evening presentations have become endowed named lectures: The Roderick, the McKusick; and the Chai lectures, respectively. These honor, Thomas Roderick, Ph.D., long time JAX staff scientist and SC co-organizer; Victor McKusick, M.D., SC founder and long-time co-organizer; and C.K. and L.C. Chai, long time JAX staff scientist and research associate, respectively, mentors to many JAX faculty and students.

Over the two weeks, the flow of the SC content is from basic human and mouse genetics topics in week one to more clinical subjects in week two. An on-site Medical Genetics clinic and a Mouse Genetics clinic take place on the afternoons of Wednesday and Thursday in the second week (see Special Features). These give the students a chance to see first-hand a wide variety of human and mouse phenotypes and how experts characterize the phenotypes and, in the case of the mouse clinic, use them to understand biology and



FIGURE 2 Photographs taken by Victor McKusick from his chair in the JAX auditorium, all with similar composition. The lecturers, from top to bottom, are Doug Coleman of the JAX, Jeff Friedman from Rockefeller, and Frank Ruddle from Yale

disease mechanisms while, in the case of the Medical clinic, how individuals with these disorders are counseled and treated.

Over the first 60 years of the SC students and faculty were all on-site, providing in-person, highly interactive experiences. In 2020, however, in the midst of the SARS-CoV-2 pandemic, the course was all virtual. This change in format reduced the opportunities for interpersonal interactions but the overall content and structure of the course was conserved. There was a major positive effect of the virtual

format on the number students and their world-wide locations (see below).

4 | ORGANIZERS

Victor McKusick was a co-organizer for the first 48 years of the SC from its inception until his death on the second day of the 48th SC in 2008. Over these years, he had a cast of fellow co-organizers (Table 1) most notably Tom Roderick, Ph.D., Staff Scientist at the JAX who served in this function for 16 years from 1974–1990. Tom and his wife, Hilda, were good friends of Anne and Victor and they thoroughly enjoyed working together on the SC. In addition to his organizational efforts and knowledge of mouse genetics, Tom brought many things to the Short Course including the idea to recognize lecturers who made outstanding contributions to the course with a carved wooden mouse, a “Roscoe,” which was awarded with much fanfare in an evening session (Figure 3). Tom and Hilda also instituted a mid-course dinner at their house, for the organizers, the Senior Lecturer

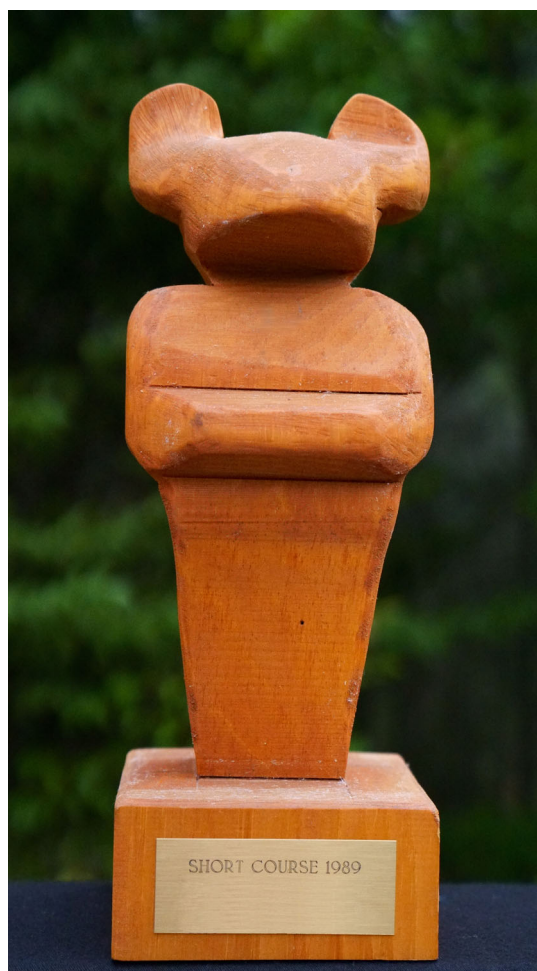


FIGURE 3 A “Roscoe”, awarded each year to a faculty member who has made outstanding contributions to the SC. The Roscoe awardee for 2021 was John A. Phillips, M.D., a long-time member of the faculty who has contributed in many ways to the success of the SC

and others, known as the “Ice Cream Social” in recognition of the tradition of hand-cranking home-made ice cream for dessert (see Figure 4).

In recent years, the task of organizing the Short Course has involved a larger team (see Table 1) who meet by phone at two-week intervals starting in September to plan the next year’s course. This effort is greatly facilitated by administrative support from the lab provided in recent years by Erin McDevitt and Katie Hodgkins.

5 | FUNDING

Over the years, the SC has been funded, primarily, by the March of Dimes and the NIH. From its inception in 1960 until 2018, grants from the March of Dimes (previously The National Foundation) supported the SC. In 1985, JAX received T15 grant support from the National Institute of Child Health and Development (NICHD). Five JAX faculty have served as PIs on NICHD course support grants from 1985 to present; during the last decade the mechanism shifted from a T15 to a R25. Greg Cox is PI with David Valle as co-PI on the current award (5R25HDO079344-08). The SC has also been supported by an Advanced Courses grant from the Howard Hughes Medical Institute (HHMI) and receives registration funds from participants. Over the last decade significant student scholarship funds have been provided by HHMI and NICHD. Details regarding the registration fees and scholarships are available at the SC web site, <https://www.jax.org/education-and-learning/education-calendar/2021/07-july/62nd-mckusick-short-course>.

6 | PARTICIPANTS

Students – Over the first 61 years, there have been 7053 students, 6084 in-person through 2019 and an additional 969 virtual in 2020. Although the majority come from North America, every continent save Antarctica, has sent students. Many have taken the course multiple times.

Faculty – There have been a total of 632 faculty from 208 institutions located in at least 15 countries in at least four continents (Africa, North America, Europe, and Australia) (see Appendix S3). The institutions sending the largest fraction of faculty include the JAX (121, 19.2%); JHU (109, 17.3%); Harvard/Broad/MIT (39, 6.2%); NIH (43, 6.8%); University of Pennsylvania (14, 2.2%); and Baylor College of Medicine (11, 1.7%). At least 28 Short Course students have returned as faculty.

The faculty are chosen on the basis of their expertise in an area of genetics, their ability as effective lecturers and teachers and their willingness to interact with students and participate fully in the course. The quality of the faculty is impressive. Over the 61 years of the SC, 17 or 2.7% of the faculty have been or subsequently were recipients of the Noble Prize in Medicine or Physiology or a related discipline (Table 2). Another 41 or 6.5% of the faculty were or subsequently were recipients of the Allan Award, the highest recognition given by the American Society of Human Genetics (ASHG) for

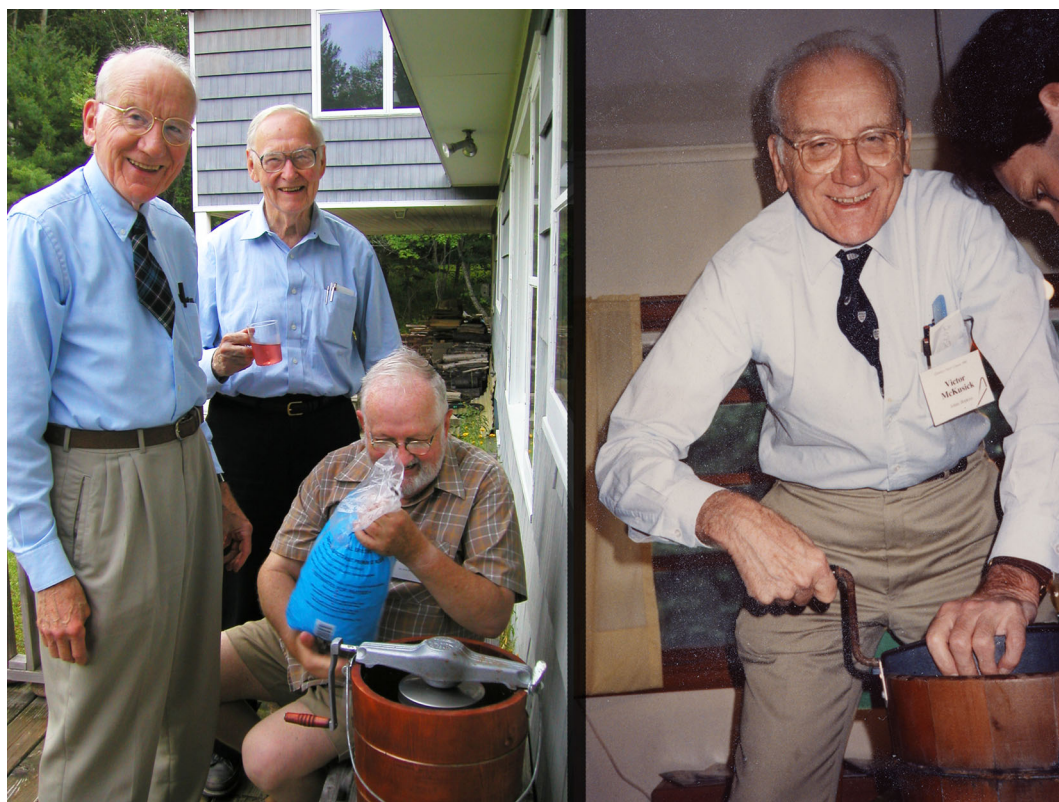


FIGURE 4 The “ice cream social” at the home of Hilda and Tom Roderick. On the left, Victor McKusick and Jim Crow watch Tom Roderick load salt into the ice cream maker; on the right, Victor does his turn cranking

TABLE 2 Nobel laureates who lectured at the course

Nobel prize year	Name	Short course lecturer year
2003	Peter Agre	2004
2004	Richard Axel	2009
1975	David Baltimore	1984
2002	Sydney Brenner	2001
1985	Michael Brown	1981
2007	Mario Capecchi	1990–95, 2009
2007	Sir Martin J. Evans	1985
1985	Joseph Goldstein	1974, 1994
2009	Carol Greider	2016, 2018
1978	Daniel Nathans	1984
2019	Gregg Semenza	1990–2021
1993	Phillip Sharp	1987–88, 2015
2007	Oliver Smithies	1996
1980	George Snell	1961–1981
2009	Jack Szostak	2018
1987	Susumu Tonegawa	1983–1984
1989	Harold Varmus	1999, 2004, 2017

“substantial and far-reaching scientific contributions to human genetics” (Table 3). Many of the Nobel Laureates and Allan awardees have lectured multiple times.

7 | CONTENT

Not surprisingly, the content of the SC has evolved dramatically over the last 60 years, reflecting the rapid advances in mammalian genetics and genomics and the growth of medical genetics over that interval. Imagine, for example, the mix of wonder and bewilderment if it were possible to fast forward an expert SC faculty member from the 1960s to the SC of 2021. DNA sequencing, epigenetic modification, mRNA splicing, SNPs, GWAS, gene therapy, the plethora of molecularly defined monogenic diseases, PCR, genome editing, transgenic mice, induced pluripotent stem cells, CRISPR and much more would all be new concepts, methods and opportunities! A thorough review of the evolution of the SC content would be an accounting of the advances in genetics over this time frame and is beyond the scope of this short paper. To give some flavor of the progress, we will mention a few highlights by decade along with a few of the speakers associated with these advances.

In the first 10 years, the content of the SC first week was dominated by discussions of the chemistry of DNA (Howard Dintzis), statistical methods for analyzing segregation and linkage (Edmund Murphy, Helen Abby, Newton Morton, Earl Green), protein biochemistry and variants (Samuel Boyer, Harry Harris), and aspects of transplantation genetics mainly in mouse (George Snell). Biochemical genetics and inborn errors of metabolism were also covered (James Sidbury, Barton Childs, James Wyngaarden) and, perhaps more than

TABLE 3 American Society of Human Genetics Allan Awardees who have lectured at the short course

Allan award year	Name	Short course lecturer number of years
1962	Newton Morton	1
1965	James Neel	1
1968	Harry Harris	2
1969	Jerome Lejune	1
1970	Arno Motulsky	3
1973	Barton Childs	7
1974	Curt Stern	1
1977	Victor McKusick	44
1978	Charles Scriver	6
1979	F. Clarke Fraser	3
1980	Walter Bodmer	2
1982	Elizabeth Neufeld	2
1983	Frank Ruddle	12
1985	Joseph Goldstein	2
1985	Michael Brown	1
1987	Luca Cavalli-Sforza	1
1987	David Botstein	3
1989	Raymond White	2
1991	Janet Rowley	1
1991	Alfred Knudson Jr	2
1994	Douglas Wallace	26
1995	Kurt Hirschhorn	3
1997	Philip Leder	1
1998	Bert Vogelstein	1
1999	Stephen Warren	2
2002	Albert de la Chapelle	1
2003	David Weatherall	2
2004	Louis Kunkel	1
2005	Francis Collins	2
2007	Arthur Beaudet	1
2008	Haig Kazazian	17
2009	Huntington Willard	3
2010	Jurg Ott	6
2012	Uta Francke	5
2013	Aravinda Chakravarti	27
2014	Stuart Orkin	8
2015	Kay Davies	2
2017	Keri Stefansson	1
2018	Eric Lander	9
2019	Stylianos Antonarakis	37
2020	Mary Claire King	6

any other subject, have remained a constant topic in the SC to the present as more and more disorders have been recognized and defined at the molecular level. The second week of the SC in these

early days focused on various genetic disorders, their phenotypes and inheritance.

By the start of the second decade of the SC (1970s), cytogenetics had become a more prominent topic with the development of banding technologies, making it possible to detect and characterize structural aberrations and to explore sex determination (Mary Lyon, Jack Miller, John Hamerton, Barbara Migeon, Bruce Cattanach). The 70s also saw the growth of somatic cell genetics contributing to gene mapping and to biochemical genetics (Frank Ruddle, John Littlefield, Liz Neufeld). Biochemical genetics continued to be prominent (Rodney Howell, Leon Rosenberg, Charles Scriver, George Thomas, Joe Goldstein, Mike Brown). Lectures on manipulating the mouse genome began to appear (Leroy Stevens) and on mouse models with high relevance to human disease (Doug Coleman). Also, the 70s saw the advent of molecular biology as a major subject with the first lectures on restriction endonucleases and their applications and molecular cloning and mapping (Daniel Nathans.).

Molecular biology became a dominant subject in the 80s. The use of restriction fragment length polymorphisms (RFLPs) as markers for mapping was first proposed in a talk by David Botstein given in the SC of 1979 and published in the seminal Botstein, White, Skolnick and Davis paper in 1980 (Botstein et al., 1980). In rapid succession, the value of associating various RFLP haplotypes with specific hemoglobin mutations was described (Stuart Orkin, Haig Kazazian, Stylianos Antonarakis) (Orkin et al., 1982) followed in 1985 by the discovery and application of the polymerase chain reaction (PCR) discussed at the SC by Norman Arnheim, Haig Kazazian and many others (Saiki et al., 1985). DNA sequencing and its applications also became a significant SC topic discussed by Lee Hood, Francis Collins, Eric Lander and many others. The availability of these molecular tools lead to the rapid and continuing increase in Mendelian disease gene discovery discussed over many years to the present, most notably by Stylianos Antonarakis. Population genetics was discussed at a largely theoretical level by James Crow and others.

In the 90s, the power of big data strategies became subjects of many SC lectures. Followed in the 2000s by completion of the Human Genome Project (HGP) adding to our understanding of the content and organization of the human and mouse genomes and recognition of the many experimental approaches made available by these developments (Francis Collins, Eric Lander). Over this period, topics that had been prominent in the early years, statistical methods, cytogenetics, tissue histocompatibility and molecular methods, increasingly became integrated into many lectures and were no longer included as stand-alone lectures. Similarly, the emphasis on gene mapping subsided as the HGP provided a nearly complete map for all genes (The International Human Genome Mapping Consortium, 2001; Venter & The Celera Genomics Sequencing Team, 2001). Mouse models became increasingly sophisticated with greatly expanded use of inbred strains as well as the introduction and development of collaborative cross and diversity outbred stains for mapping and identifying genes involved in complex traits (Churchill et al., 2004, 2012). Over this same time frame, the second week of the SC continued to focus

on clinical topics but, in contrast to the earlier years, these included descriptions of the responsible genes and variants. Similarly, population genetics increasingly incorporated molecular data and the first genome wide association studies (GWAS) began to be published and to be discussed in the SC (David Altshuler, Aravinda Chakravarti, Goncalo Abecasis) as well as progress in using these tools to understand the genetic architecture of common complex traits (Peter Visscher, Judy Cho and many others). Topics with Ethical, Legal, and Social Implications (ELSI), first discussed in the 90s, occupied a larger fraction of the SC program as sequencing and the application of big data approaches expanded. These technologies not only provide increased insight to disease but also expand opportunities for misuse of genetic data, necessitating careful consideration of ELSI principles and practices.

The last decade, has seen a plethora of new topics appear in the SC including: genome organization and packing, advances in sequencing especially in long-read methods, epigenetic mechanisms and diseases, regulation of gene expression, genetic architecture of complex traits, polygenic risk scores, continued growth of gene discovery and pathologic mechanisms for rare Mendelian disease, increasing ELSI challenges, and, gratifyingly, a noticeable increase in the development and application of effective treatments for genetic disease. The increasing integration of genetics and genomics into medical practice and thinking has been a common theme for the clinical topics. The pace of these advances and discoveries continues to increase; one can only imagine with excitement what the next years will hold.

8 | SOME OTHER SPECIAL FEATURES

In addition to lectures and workshops, there are several special features which have added value to the SC experience.

8.1 | Medical clinic

From the beginning, the SC has included afternoon medical genetics clinic, held in the second week and typically staffed by SC faculty and students who see 5–10 patients and their families which are then discussed at a post clinic conference open to all students. Initially, this effort was led by Victor McKusick with help from Reed Pyeritz, Clair Francomano and Judy Hall. More recently, Ada Hamosh with the help of many residents in training, organize this effort. The patients come from Maine and surrounding states and, for many years, many were identified and recruited by the Center for Human Genetics, a small clinic headed up by Melba Wilson, located in Bar Harbor. The patients take advantage of the opportunity to consult with experienced medical geneticists while the SC students have the opportunity to observe a medical genetics clinic in action: the kinds of patients, their management and their concerns, as well as genetic counseling issues. For many SC students, this is their first exposure to patients with genetic diseases.

8.2 | Mouse clinic

Also from the beginning, a mouse mutant “clinic” has been a tradition occurring on an afternoon in the second week. Inbred mouse strains demonstrating the phenotypes of various mutations are demonstrated by JAX scientists and mouse handlers. In 2019, the mouse clinic was named after long-time Research Associate Priscilla “Skippy” Lane to honor her 56-year career at JAX that included presenting mutant mice in the SC every year until 2007. Typically, 50 or more mutant mouse strains are available grouped by affected system (hematologic, skeletal, endocrine, etc.) including many classic mouse mutant strains (Ob, Tubby, NSG, DBA, etc.) as well as newly recognized spontaneous mutant strains yet to be characterized. The students can observe these models “on-the hoof” and can discuss the phenotypes with experienced mouse biologists. This exposure provides a valuable opportunity to appreciate the breadth and diversity of mutant mouse strains and the opportunities they provide for modeling human disease.

8.3 | ELSI night

Genetic medicine touches many ethical, legal and social issues (ELSI). These are discussed in one or two lectures in the second week but to consider and discuss real life scenarios, the SC has for many years had an ELSI evening led by four faculty members who represent the key principles of ethical decision making including in the early years: beneficence, Clair Francomano; nonmaleficence, Laird Jackson; autonomy, John Phillips; and justice, Judy Hall. Real examples are presented and discussed guided by the four with, in recent years, commentary from geneticist and bioethicist Debra Mathews. The diversity of viewpoints advocated by the students emphasize the complexity of these issues, the factors that should be considered and the challenge of weighing all the variables to reach a reasonable and fair decision. Based on the student turnout and lively discussions, the organizers concluded this was the most effective way of exposing and engaging the students to the complexity and value of an ethical framework for ELSI issues.

8.4 | Press week

In the early 1960s, not long after the SC got started, Victor and Alan Davis, a JAX trustee with connections to the American Cancer Society, conceived of inviting members of the national science press to attend one week of the SC. This gave the press an opportunity to hear the progress in genetics and to interview leaders in the field. Their efforts lead to publicizing the SC and educating the public on the progress of genetics research and its contributions to medicine. Stories with the byline, “from Bar Harbor, Maine” appeared in major publications including the New York Times, the Wall Street Journal, Newsweek and many others. Connections made between writers and scientists during Press Week often led to communications throughout

the remainder of the year for stories and comments. At its peak, 20–30 members of the science press participated in Press Week. Over the last few years, however, as budget restrictions and the use of electronic communication increased, Press Week attendance fell and this tradition has now become a thing of the past.

8.5 | Laird Jackson and the SC tee shirts

Laird Jackson, M.D., was a student at the SC in 1961 and returned nearly every year thereafter as a member of the faculty until his death in 2019, speaking on topics related to clinical genetics, prenatal diagnosis and genetic counseling. Laird conceived of and for more than 20 years supplied a treasured SC memento for all students and faculty, an annual SC tee shirt. A different bright color each year, the tee shirts were decorated front and back with cartoons drawn by Laird and depicting key points from important publications from the previous year (see Figure 5).

8.6 | Anniversary celebrations

At roughly 10 year intervals, special celebrations were held to mark the progress of the SC. The 40th was particularly notable with a lobster dinner under the stars accompanied by Francis Collins singing a special composition, “Never Say Vic” (see Appendix S2).

9 | THE IMPACT OF THE SHORT COURSE

Without question, the SC has vastly exceeded McKusick and Fuller's original goal of “teaching the teachers.” Over six decades, nearly 8000 students and faculty have participated in the course receiving an intense, ~8 h/day, two-week exposure to the state of the art of genetics at the time. The participants, faculty and students alike, experience, first hand, good science communication from lecturer after lecturer. They have the opportunity to talk informally with the speakers over coffee or lunch or on a hike in Acadia National Park to get questions answered and discuss science and career plans. Participants also receive a comprehensive set of notes, outlines, references and, more recently, slide decks that many use thereafter as guides for their own educational activities. The development of “satellite” courses modeled after the SC has served to extend the educational impact of the Bar Harbor course to students in Europe and in China (see Romeo et al., 2021).

The impact of the SC on science and research in genetics has also been considerable but is difficult to quantify. SC participants are exposed to high quality genetic and genomic research applying the latest technologies to answer important questions. The faculty are selected for the quality of their science as well as their abilities as good communicators. The informality of the SC provides many opportunities for discussions about the latest science with leaders in the field.

Moreover, the comprehensive scope of the SC content is sure to expose those working in one area to what is going on in others. Emphasis on the mouse/human synergisms and the contributions of

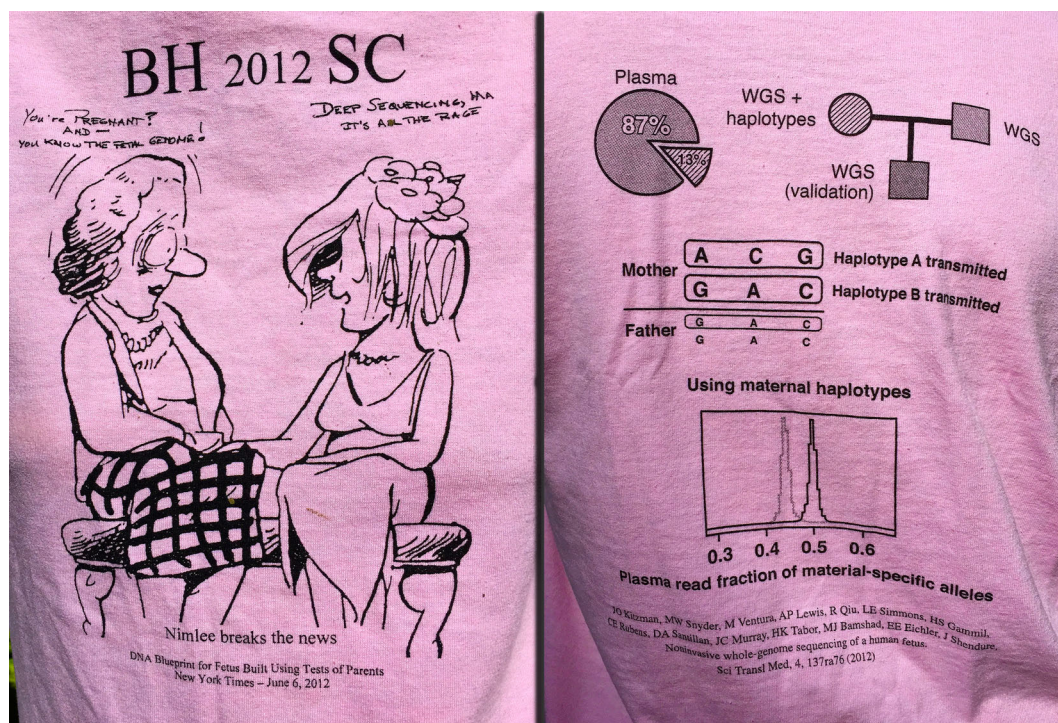


FIGURE 5 The Short Course Tee Shirt from 2012 designed, as always, by Laird Jackson. The front (left) shows a pregnant woman telling her mother that the genome of her fetus has been sequenced; the back (right) shows data from the paper published by Evan Eichler and Jay Shendure and their colleagues

the mouse as a model for human genetic disease has also influenced the research directions of many participants. These factors and others explain the oft repeated comment “I got this idea at the SC.”

9.1 | The impact on Johns Hopkins (JH)

In addition to the 106 JH faculty members who have lectured at the SC (from 1 to 46 years each) over the six decades of the SC, we send our trainees (graduate students, residents, fellows, roughly 15–20/year). Many of our non-geneticist Hopkins colleagues (5–10/yr) opt to take the course as students for the opportunity for an intense exposure to genetics in a beautiful environment.

The value of teaching at the SC has been an important component in our faculty's development as educators learning how to effectively communicate their science. The experience also makes us better geneticists; to paraphrase an old axiom, “the teacher is the one who gets the most out of teaching.”

For those faculty participating year after year, the SC also provides an intense annual refresher—a snapshot (or really a panoramic image) of the state of the field. The value of this is difficult to quantify but is considerable. For example, the senior author of this article by the time of publication will have had the benefit of attending 45 years or 90 weeks (1.7 years) of the SC in two week doses, learning what is new, re-visiting difficult topics and interacting with colleagues; what a privilege!

As mentioned, for the last four decades, the JH Human Genetics graduate students (about 12/year) attend the SC in the

summer between their first and second year. This experience repeats what they may (or may not) have learned in their formal first year course work, provides a deep exposure to mouse genetics and the value of the mouse as an experimental system and helps then establish connections with other students of their generation which often last for the entirety of their careers. At their PhD thesis seminar marking the end of their graduate experience, virtually every one of our students reminisces about their SC experience.

9.2 | The impact for the JAX

The benefits for JH described above all also apply to JAX. Additionally, and particularly in the early years, the SC was a major window of exposure of the JAX and JAX scientists to geneticists working outside of Bar Harbor, Maine. The SC gave the JAX faculty and trainees a chance to meet geneticists and trainees from around the country, especially those with expertise in human disorders seeking to model their favorite phenotype in an experimentally relevant and tractable model organism such as the mouse. This networking value, first exemplified by the SC, has been recognized and embraced by JAX; one needs only to look at their current extensive line up of >20 courses and conferences which not only educate but also bring a steady flow of outside scientists and physicians to JAX. (<https://www.jax.org/education-and-learning/course-and-conferences>).



FIGURE 6 Class Photograph for the 47th Annual Short Course, 2006, the last year Dr. McKusick was able to attend in person. Participants in the first row from left to right include: Jennifer Puck, M.D.; Robert Nussbaum, M.D.; Katherine High, MD; Philip Reilly, MD.; Unknown; John Phillips, M.D.; Roderick McInnes, M.D.; Victor McKusick, M.D.; Patsy Nishina, Ph.D.; Jurgen Naggert, Ph.D.; David Valle, M.D.; Ada Hamosh, M.D.; Clair Francomano, M.D.; Garry Cutting, M.D.; Hal Dietz, M.D.; Unknown; Unknown

10 | THE FUTURE

The content of the SC will continue to evolve and provide exciting new information for geneticists of the future. We are constantly evaluating the SC format. We viewed the challenges of the SARS-CoV-2 pandemic with trepidation, but by all accounts, the all virtual 2020 SC was a qualified success. The all-virtual venue allowed us to reduce costs and provide student access for a very reduced fee. As a consequence, the number of students increased nearly 10-fold to 969 students from 6 continents. We missed, however, the direct interpersonal interactions of the all on-site course. We expect, going forward, that we will offer a “hybrid course” with both in-person and virtual students. We are also in the third year of developing a robust connection with the African Society of Human Genetics and geneticists across the vast African continent. In 2021, this will feature a two-hour session with speakers from Africa and a panel discussion on the challenges and opportunities of genetic research and clinical work in Africa, the birthplace of our species. We expect this model will flourish and plan to develop similar connections with geneticists in other parts of the world.

11 | SOME CONCLUDING REMARKS

The SC and OMIM (Hamosh et al., 2021) are two of the most significant contributions of Victor McKusick to medical genetics. Both have touched nearly all in our field and both keep giving now more than 10 years after his death. Both exemplify the great value of education for the student and the teacher. The organizers are committed to fulfill Victor's evaluation, made at the end of each SC, of “best yet.”

Victor loved the SC and being in Maine (Figure 6). He was noticeably more relaxed and informal in Bar Harbor than back home in Baltimore. After a long day of morning lectures, afternoon workshops and the evening lecture, Victor and the senior author of this article often shared a late-night ride back to our rental cottages on the western side of the island. On one such ride more than 20 years ago, Victor spontaneously volunteered that his goal was to be “on the podium” in his 90th year (2011). Sadly, that was not to be; he died during the SC on 22 July 2008, at the age of 86. But his intellect, spirit and enthusiasm persist to this day, so, in fact, he was on the podium on his 90th just as he was on this, his 100th, birthday.

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CONFLICT OF INTEREST

There are no conflicts of interest for the authors of this article to declare.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

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SUPPORTING INFORMATION

Additional supporting information may be found in the online version of the article at the publisher's website.

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