American Journal of Medical Genetics 72:191–196 (1997)

Association of Professors of Human or Medical Genetics: Second Annual Workshop Summary

J.M. Friedman*
Department of Medical Genetics, University of British Columbia, Vancouver, Canada

INTRODUCTION

The Association of Professors of Human or Medical Genetics held its second annual workshop at the Westgate Lakes Resort in Orlando, Florida, on 20–22 January 1996. Seventy participants representing 55 universities throughout North America attended.

Eighty-five institutions are now official members of the association, which was formally incorporated as a non-profit organization on 21 August 1995. Bob Desnick became president of the association on 1 July 1996. Jan Friedman became Past-President and Skip Elsas became President-Elect at that time. Hunt Willard and Uta Francke became councillors, replacing Jewell Ward and Skip Elsas.

On the basis of discussions held at the workshop, the council proposed the following areas of focus for the association's activities for the next year:

1. Continue to work to improve medical genetics coverage in the United States Medical Licensing Examination (USMLE) and encourage the reporting of a Step 1 genetics score to deans.
2. Implement the association's question database and provide access to participating institutions.
3. Encourage the use and development of computer-based resources for human genetics teaching.
4. Establish or enhance relationships of the Association of Professors with other organizations with complementary areas of interest, including the American Society of Human Genetics (ASHG) Information and Education Committee, the American Board of Medical Genetics (ABMG), the American College of Medical Genetics (ACMG), and the Congress of Regional Genetics Networks.

The program of the second annual workshop included four sessions, which are summarized below.

SESSION 1: EDUCATION AND EDUCATIONAL TOOLS *(Peggy McGovern, Mt. Sinai, and Bruce Dowton, Washington University, Co-chairs)*

Survey of Genetics Teaching at Medical Schools

Peggy McGovern (Mt. Sinai) described a survey she conducted of member institutions. The survey was sent to 62 institutions, of which 30 replied. Twenty-three of these 30 schools include genetics in their medical school curriculum. Three fourths of the schools that include genetics have a separate course; at the other schools genetics is integrated into one or more other courses.

The amount of time in the curriculum devoted to genetics varied from 10 to 75 contact hours, with an average of 31 hours. About one half of the courses included some element of problem-based learning (PBL), and about one half of the rest were in the process of developing PBL materials. Only 15% use computer-assisted instruction (CAI) in teaching genetics to medical students. However, 45% of the schools responding use computer resources to teach genetics to other students or trainees. The *London Dysmorphology Database®*, *POSSUM®*, *OSSUM®*, *Silver Platter: Nature of Genes®*, and *OMIM®* were used most often.

Using PBL in Medical Genetics

Ron Davidson (McMaster University, Hospital for Sick Children) discussed the development of PBL cases in genetics for medical students. Many medical schools are now adopting PBL because the rapid increase in biomedical knowledge makes life-long learning essential. PBL also takes into account differences in student background and learning styles, teaches group function, helps students learn how to use resources efficiently, integrates multiple areas of knowledge, and encourages forward reasoning.

PBL requires medical school faculty to abandon some of their traditional assumptions regarding how medical students should be taught. Tutors must be trained in the techniques of PBL and must act as facilitators of student learning, rather than as a source of information. Students should be directed toward useful resources, but not spoon fed.

*Correspondence to: J.M. Friedman, M.D., Ph.D., UBC Department of Medical Genetics, BC Children's Hospital, 4500 Oak Street, Vancouver, Canada V6H 3N1.

Received 16 September 1996; Accepted 10 February 1997

The first step in developing a PBL case is establishing the learning objectives. The case scenario is then written to bring out those objectives. Questions may be included to help direct the students toward the issues of concern. Separate student and tutor guides are then developed. The tutor guides provide background that is necessary for tutors who are not experts in the problem's content area.

Reviews of Medical Genetics Text Books

Joe Christian (Indiana University) described his experience using the Thompson and Thompson book [Thompson et al., 1991] for a medical student course. The book is somewhat out of date, but a new edition is expected this year. The text is well written and readable. It includes a reasonable amount of information for a medical student course in genetics, but the approach is not as clinical as that taken by some other books.

Greg Grabowski (University of Cincinnati) reviewed the Gelehrter and Collins book [1990]. This book is relatively short but works well for genetics courses that must be taught in a limited amount of time. The text is readable but lacks detail in some areas. The questions are generally well focused.

Marian Rivas (University of Tennessee) discussed the Mueller, Young, and Emery [1995] and the Jorde, Carey, and White [1995] books. Both are recent (1995), but they take very different approaches to presenting the material. The Emery et al. book uses a classical approach, presenting principles first and then the clinical applications. This separation of the normal and abnormal aspects of a topic often seems artificial. The book does not include problems, but it is attractively designed.

The Jorde et al. book is more condensed and stresses problem-solving, genetic approaches to disease, and genetic variation. Genetic themes are integrated throughout the text rather than being presented only in one place. Topics such as cancer genetics, common diseases, multifactorial inheritance, and mitochondrial genetics are presented well, but other topics (e.g., developmental genetics and pharmacogenetics) are not included at all.

CAI in Medical Genetics

Bruce Dowton (Washington University) demonstrated a prototype computer-assisted program he developed for teaching medical genetics. The program is designed so that it can be used by students and trainees at several different levels as well as for continuing education. The program takes into account the varying background of both students and faculty with respect to genetics and computer literacy. The prototype, which is called "Tools for Teaching Genetics," includes core concepts, skills, technologies, disease descriptions, and images as well as case studies and problem sets. These elements are assembled into an electronic textbook that can be viewed at varying levels of detail and includes hypertext links to related concepts elsewhere in the system. The software runs as a free-standing application on many different kinds of computers.

Medical Genetics Question Bank

Peggy McGovern (Mt. Sinai) has assembled a database of medical genetics questions over the past year. Twenty-four member institutions of the Association of Professors have submitted some 9,000 questions (mostly multiple-choice) from medical student exams given over the past 3 years. These questions and their associated images have been scanned into a database and divided into 6 subject categories: general genetics, cytogenetics, biochemical genetics, molecular genetics, clinical genetics, and population genetics. Further subclassification of the questions is planned.

Dr. McGovern proposed that this database be used as the nucleus of an ongoing service that would be available to members of the Association of Professors. In order to gain access to the questions, a member would submit 3 recent examinations and provide a certain number of additional questions each year. There would be a one-time administrative fee and a smaller annual subscription fee. The exact conditions for membership would be defined in consultation with the participants. The database would be distributed on disk to help preserve confidentiality. Mt. Sinai School of Medicine will serve as the coordinating center for this database.

SESSION 2: EMPOWERING GENETICS AS A DISCIPLINE WITHIN MEDICAL SCHOOLS

(Jan Friedman, University of British Columbia, Chair; organized with Jewell Ward, University of Tennessee)

Review of United States Medical Licensing Examination (USMLE) Questions in Medical Genetics

A delegation representing the Association of Professors and the American Society of Human Genetics (ASHG) visited the National Board of Medical Examiners on 4–5 December 1995 to review the genetics content of the USMLE. The delegation consisted of Jan Friedman (University of British Columbia), Skip Elsas (Emory University), Mimi Blitzer (University of Maryland), and Maimon Cohen (University of Maryland). The delegates reviewed all of the questions and answers to the most recently administered editions of the USMLE Step 1, Step 2, and Step 3 examinations. Each Step examination includes 720–730 multiple-choice questions that had been administered over 2 days.

After reviewing the examinations, the delegates made both qualitative and quantitative observations. These include:

1. Less than 5% of the items asked questions requiring knowledge of genetics. Of these items, only about one third dealt with important genetic principles; the other items tested specific facts related to individual genetic conditions.
2. More than two thirds of the genetic items were on the Step 1 examination. There were very few genetic items on either Step 2 or Step 3, and almost all of these tested specific facts related to individual genetic diseases rather than important principles of medical genetics.

3. The distribution of items within the field of genetics was skewed, probably reflecting the structure of the Test Materials Development Committees that put items on the examinations. Some areas were reasonably well covered. Other areas were covered poorly or not at all.
4. Several items had "correct answers" that the delegates considered to be wrong because current genetic understanding was ignored.
5. A positive family history was included as extraneous information in some questions. There were other instances in which the list of answers included one or more genetic diseases as incorrect responses.
6. There were many missed opportunities to test important genetic principles. This usually could have been achieved by changing just a few words in the question or responses. There were also several instances in which a question about microbial genetics could have been made clinically relevant by asking the question about human genetics instead.

After reviewing the USMLE test materials, the delegation met with Dr. L. Thompson Bowles, President of the National Board of Medical Examiners, Dr. Donald E. Melnick, Senior Vice President, and their senior staff who are responsible for the USMLE Step 1, Step 2, and Step 3 examinations. The delegates discussed their findings and concerns about the genetics content of the examinations.

The delegates were disappointed to learn that there are currently only 3 or 4 human geneticists on any of the USMLE Test Materials Development Committees or Step Committees. The delegates believe that the medical genetics coverage of the USMLE could be improved substantially if more individuals with expertise in this area took part in writing, reviewing, and selecting questions for the USMLE. The delegation, therefore, urged the National Board of Medical Examiners to establish a Medical Genetics Test Materials Development Committee and to appoint qualified medical geneticists to each of the Step Committees.

As teachers of medical genetics, the delegates felt that it would be very useful for the USMLE to report a genetics subscore on the Step 1 examination to the medical school deans in the same way that subscores in other subject areas are reported. The officers and staff of the National Board of Medical Examiners received these comments with great interest and will take the recommendations made to the various constituent committees for consideration.

[*This report has been approved for release by the National Board of Medical Examiners.*]

Improving Clinical Genetics Research

Jim Hanson (National Institute of Child Health and Human Development [NICHD]) reviewed how health care reform, in general, and Medicare reform, in particular, threaten the financial foundations of academic medical centers. Most genetic research sponsored by the U.S. government is funded through the National Institutes of Health (NIH). Less than one half of this funding goes to the National Center for Human Genome Research. Other funds flow through various institutes including the NICHD, the National Cancer Institute (NCI), the National Institute of Environmental Health Sciences (NIEHS), and the National Institute of General Medical Sciences (NIGMS), among others. Most of the support goes to basic science research; only a small amount is spent on clinical research. There is no coordination of clinical genetics research activities either within the NIH or between the NIH and other agencies involved (e.g., the Centers for Disease Control and Prevention [CDC] and the Health Resources and Services Administration [HRSA]).

Since 1986, the NICHD has funded clinical research networks. Such networks are currently in operation in the fields of neonatology, perinatology, and pediatric pharmacology and drug development. These networks function as collaborative agreements between the NIH and academic clinical centers that are chosen competitively. Once established, the network identifies research problems, prioritizes them, and carries out appropriate studies. The networks have focused largely on clinical trials, although some observational studies and other investigations have been pursued. Dr. Hanson believes that the NICHD would consider the development of a clinical genetics network, although the value of such an enterprise would have to be very clearly demonstrated.

Creating a Medical Genetics Department in the Academic Medical Center

Kurt Hirschhorn (Mt. Sinai) discussed what is necessary to create an academic medical genetics department in a center that does not currently have one. The factors include:

1. High-quality funded research.
2. Wide recognition of the need for a medical genetics department outside the medical genetics unit.
3. Convincing the dean and university administration that excellence in medical genetics is important to the future of the medical school as a whole.
4. Building support for a medical genetics department among the medical school department heads.
5. Working out overlaps in research, teaching, and patient care.
6. Finding a powerful spokesman at the senior level of medical school decision-making.

The specific approach taken will vary from institution to institution, depending on local considerations.

SESSION 3: CLINICAL GENETICS TRAINING PROGRAMS *(John Carey, University of Utah, and Skip Elsas, Emory University, Co-chairs).*

John Carey (University of Utah) summarized the existing American Board of Medical Genetics (ABMG) requirements for clinical genetics fellowships and compared these requirements to those of the Medical Genetics Residency Review Committee (RRC). The RRC guidelines are more specific with respect to issues such as the amount of time devoted to clinically oriented activities, the evaluation process for residents, and the amount of weekend and night call permitted.

All clinical genetics training programs in the United States will require accreditation by the RRC by 1 January 1998. After that time, the ABMG will no longer accredit clinical genetics programs, but will continue to accredit training programs in genetic laboratory specialties. However, the ABMG will require a center to have an RRC-accredited clinical genetics training program in order to be considered for accreditation of genetic laboratory training programs.

Study of ABMG Training Program Applications

Dr. Carey presented a study of ABMG clinical genetics training program applications received between 1991 and 1993. The analysis was based on the proposed weekly schedules (Part XVI) which were complete enough for study in 54 (78%) of the 70 applications. Clinical fellows were expected to take part in an average of 4 half-day genetics clinics per week in Year 1. Fellows were expected to participate in fewer clinics during the second year in 65% of programs. Most programs include 1 or 2 specialty clinics per week; 26% of programs have no prenatal diagnosis clinic on their schedules. Clinical laboratory experiences vary greatly. Over 90% of the programs have 4 or more hours per week of conferences, seminars, and rounds. Three fourths of the programs include formal courses for the fellows. Research rotations usually occur in the second year and may last up to 6 months.

The degree to which fellows' actual schedules vary from those described is not known. The study raises questions about the appropriateness and quality of the training being provided.

Examples of Clinical Training Programs

Skip Elsas (Emory University) described the program at Emory which is accredited for all ABMG specialities. The program has a very academic focus and requires a third year of fellowship to provide research training for M.D.s who do not also hold a Ph.D. Formal course work is required, and there are rotations through the cytogenetics, biochemical genetics, and molecular genetics laboratories. Fellows spend 1–2 days per week in clinics and clinical conferences.

Bruce Korf (Boston Children's Hospital) summarized the Harvard program which involves 7 different hospitals. This ABMG-accredited clinical fellowship program requires a third year for all fellows. The first year is devoted to rotations through various clinical genetics services and laboratories. Second-year fellows attend one clinic per week and take weekend calls; the remainder of their time is spent on research. The third year is devoted entirely to research. All fellows participate in weekly conferences, rounds, and seminars.

John Carey (University of Utah) provided an overview of the University of Utah program. This is a 2-year ABMG-accredited fellowship with an optional third (research) year. All fellows take 3 formal courses in medical genetics and participate in a variety of genetics conferences each week. The fellows attend 1–2 genetics clinics per week and take part in other speciality clinics and a consult service as well.

Integration of Laboratory and Formal Genetics Experiences

Maimon Cohen (University of Maryland) discussed cytogenetics laboratory experience in the training of clinical geneticists. The goal should be to give the clinical geneticist an understanding of how the laboratory should be used in arriving at a diagnosis and in providing counseling to affected families. Laboratory training for clinical fellows should involve a didactic component and some general hands-on experience. Involvement of the trainee in interpretation and sign-out of specimens is particularly important.

Art Beaudet (Baylor College of Medicine) explained that Baylor does not require clinical fellows to rotate through genetics service laboratories, but fully incorporates the laboratories into all aspects of the fellowship. Fellows take a formal graduate course that deals largely with molecular and biochemical genetics. Laboratory data and their interpretation are essential components of weekly teaching rounds and conferences and often come up in the management of individual cases.

Walter Nance (Medical College of Virginia) asserted that knowledge of formal genetics is what separates medical geneticists from other physicians. It is, therefore, essential that all clinical genetics fellows have training in formal genetics. At the Medical College of Virginia, fellows attend an introductory graduate course in human genetics and are encouraged to take other in-depth courses in formal genetics. Formal genetics is also included in medical genetics journal clubs and seminars.

During the discussion following these presentations, it was recommended that future Association of Professors workshops consider methods of improving medical genetics training programs, a core curriculum for medical genetics residents and fellows, whether an additional year of training is necessary, and the career for which medical geneticists should be trained.

The ASHG Rapid Action Team (RAT) on Genetics Residency Training

Suzanne Cassidy summarized a recent meeting of the ASHG Rapid Action Team on funding of clinical genetics training. The RAT met in Chicago on 2–3 December 1995. Participants included Dr. Cassidy (chair), Judy Hall (ASHG), Jim Hanson (NICHD), Jessica Davis (American College of Medical Genetics [ACMG]), Gene Hoyme (ABMG, American Academy of Pediatrics), Claire Francomano (National Center for Human Genome Research), Jan Friedman (Association of Professors, Canadian College of Medical Geneticists), and Elaine Strass (ASHG). David Rimoin (ACMG) took part by conference telephone call.

The RAT identified several key issues related to the training of clinical geneticists. These issues include the role and spectrum of activity of clinical geneticists, the necessity of conducting a needs assessment for clinical geneticists, the importance of identifying sources of funding for training clinical geneticists and for clinical genetics research, and the question of whether clinical genetics will be financially viable as a specialty in the future.

In order to address these issues, the RAT began to develop a vision of the clinical geneticist as a specialist whose key role is in translating genetic science into medical practice. Conducting a needs assessment in clinical genetics was considered to be a very high priority; such a needs assessment will be undertaken by the ACMG. It was agreed that clinical genetics training should be paid for by teaching hospitals in the same manner as other kinds of medical specialist training. Few medical genetics training positions are funded in this manner at present, but a strategy is being developed to deal with this problem.

Improvement of Genetics Training Programs

In the discussion that followed these presentations, the issue of whether medical geneticists should be trained to pursue academic careers or careers that are primarily in clinical practice was touched on but not discussed in detail. This is an important question that needs to be considered in depth at future meetings.

The major thrust of the RRC training program requirements is to prepare medical geneticists for clinical practice. As the RRC replaces the ABMG as the accrediting body for medical genetics training, individuals who wish to pursue an academic career may need to obtain at least one additional year of research training.

The RRC has proposed a change in its guidelines so that on-site cytogenetics, biochemical genetics, and DNA diagnostic laboratories will no longer be required. Although on-site laboratories are desirable for training clinical geneticists, retaining all of these laboratories may be impractical. The number of comprehensive training programs that have all clinical and laboratory components available on site is likely to decrease.

This session was one of the first public discussions of structural issues related to clinical genetics training since the establishment of ABMG programs more than a decade ago. Consideration of how to improve clinical genetics training should be continued in association with the ACMG and ABMG. It was suggested that future Association of Professors workshops include reports from the ABMG and the ACMG as well as from the USMLE liaison committee and the NIH on training and research issues in medical genetics.

SESSION 4: DEFINING THE SPECIALTY OF MEDICAL GENETICS *(Bob Desnick, Mt. Sinai, and Emmanuel Shapira, Tulane University, Co-chairs)*

Bob Desnick (Mt. Sinai) reviewed the development of medical genetics as a specialty from its infancy in the 1940s, through childhood in the 1970s and 1980s, to adolescence in the 1990s. The recent (1993) acceptance of the ABMG into the American Board of Medical Specialties (ABMS) marks the beginning of full recognition of medical genetics by the American medical establishment. Nevertheless, medical genetics remains a junior partner in American medicine, being one of the smallest specialties in the ABMS.

Medical genetics is defined as a specialty by its special clinical expertise. This includes genetic counseling and the diagnosis, management, and treatment of genetic diseases and birth defects. Special laboratory testing and interpretation are also primary foci of medical genetics practice. The clinical activities of medical geneticists are closely tied to continuing advances in genetic research, and clinical geneticists play a key role in translating research advances into patient care.

Clinical geneticists have developed a relatively well-accepted role in the diagnosis and management of genetic diseases and birth defects in children and in prenatal diagnosis counseling. Their role is less generally accepted in adult medicine or for common multifactorial conditions. Some clinical geneticists serve exclusively as consultants while others are primary care providers for patients with genetic diseases or birth defects.

Health care reforms are threatening the viability of clinical genetics laboratories within academic medical centers. The clinical and academic value of these laboratories will have to be weighed against their financial costs in deciding whether they can remain a central focus of academic medical genetics units. New areas of emphasis need to be considered by clinical geneticists as they plan for the future.

Emmanuel Shapira provided an overview of some of the problems and opportunities facing genetic clinical and laboratory services. Inadequate funding is currently the biggest threat to academic medical genetics programs. Clinical genetics services often overlap with those of other specialists, which may lead to competition for patients within a medical center. Clinical laboratories, which are important for training and research as well as for generating income, now face nationwide competition from for-profit laboratories that often have access to enormous financial and infrastructural resources. In addition, HMO contracts with university teaching hospitals may require laboratory work to be sent to designated outside laboratories.

Maintaining/Expanding the Genetic "Turf"

Peggy McGovern (Mt. Sinai) discussed the role of medical geneticists as physicians who are primarily responsible for the care of patients with genetic diseases. This occurs most often in pediatrics because many clinical geneticists are pediatricians in pediatric departments. Clinical geneticists have generally played a much more limited role with respect to adult patients, although some geneticists have retained primary responsibility for adults with inborn metabolic errors. Most medical geneticists serve as consultants for patients seen by other physicians, but it is uncertain how this will evolve as more genetic services are provided to such patients.

The roles medical geneticists play in the future may in large part be determined by whether they are trained solely in medical genetics (i.e., through a 4- or 5-year medical genetics residency) or are qualified in another specialty as well as in medical genetics, as is the case for most clinical geneticists currently in practice. Establishing joint programs that will train residents in both medical genetics and another specialty such as pediatrics, internal medicine, or obstetrics and gynecology, should be explored.

Len Pinsky reported that he has served as the "conscience of genetics" at Medicine Grand Rounds at McGill for many years. In this role, Dr. Pinsky has assured that internists learn the importance of obtaining a proper family history and of using this information in their practices.

The genetics and obstetrical services have worked closely together for more than 20 years at Yale, Maurice Mahoney reported. This has been fostered by shared research and training interests, but has also depended on a rapid clinical response and appropriate expertise among the medical geneticists. The roles of medical geneticists, perinatologists, and genetic counselors in prenatal diagnosis services vary from center to center, with consequent differences in the cost, amount of counseling provided, and the overall quality of care.

Mark Evans described how clinical genetics services are integrated into the obstetrics department at Wayne State University. This has permitted development of a strong and well-coordinated program of prenatal diagnosis, counseling, and treatment. The relationship of clinical geneticists and obstetrical ultrasonographers is particularly important because of the increasing frequency with which fetal anomalies and genetic diseases are being identified by ultrasound examination.

Maimon Cohen (University of Maryland) considered the role that cytogenetics laboratories play within academic clinical genetics units. The research and teaching functions of the laboratory can be carried out most effectively if the cytogenetics and clinical genetics services are part of the same unit. However, the academic functions of the lab may make it less efficient than large commercial laboratories and threaten its financial viability. The unique expertise of clinical geneticists and cytogeneticists in interpreting cytogenetic findings needs to be exploited as a strong advantage of academic laboratories.

Herb Lubs (University of Miami) presented data collected for academic and commercial cytogenetics laboratories in the southeastern United States from 1983 to 1994. Together these laboratories perform cytogenetic analysis on about 15,000 leukocyte cultures each year; this number has not changed over the past 10 years. In contrast, the number of amniotic fluid cytogenetic studies has increased from 10,000 to 30,000 per year during that period. All of the growth has occurred in large commercial laboratories. Efficiency in terms of the number of studies completed per staff member is more than three times greater in large labs (>1,500 specimens per year) than in small labs (<500 specimens per year). Among large labs, commercial labs are 1.5–2.5 times more efficient than academic labs. Academic cytogenetics laboratories are unlikely to be financially competitive under these circumstances.

The biggest threat to academic laboratories is "full-service contracts" that require all laboratory services on covered patients to be sent to a single outside laboratory. Differences in the quality of service and the advantages of providing training may be relatively unimportant to insurers negotiating these large, comprehensive contracts. Academic laboratories will need to understand their competition and be more innovative and efficient in order to survive in this financial climate.

Skip Elsas (Emory University) pointed out that clinical biochemical genetics and molecular genetics laboratories must draw specimens from very large population areas in order to be cost efficient. In many states, public health activities such as newborn screening and follow-up of abnormal screening results have been important in supporting biochemical genetics laboratories, but privatization of government services is threatening this system.

Aubrey Milunsky (Boston University) described how academic laboratories might cooperate for their mutual benefit. This would require setting aside traditional rivalries among universities and developing the strong leadership and business infrastructure necessary to compete with corporate professionals. A first step toward development of a consortium of academic genetic service laboratories would be maintaining an up-to-date directory of the services provided by such laboratories. This could be made available over the Internet at minimal cost. To compete effectively, academic laboratories will have to maintain high standards and provide the same rapid turn-around time and service orientation as commercial labs.

Walter Nance reviewed the development and growth of the Department of Human Genetics at the Medical College of Virginia since 1975. A key factor in this success has been nurturing effective communication and a supportive relationship with both the medical school dean and the director of the teaching hospital.

REFERENCES

Gelehrter TD, Collins FS (1990): "Principles of Medical Genetics." Baltimore: Williams & Wilkins.

Jorde LB, Carey JC, White RL (1995): "Medical Genetics," St. Louis: Mosby.

Mueller RF, Young ID, Emery AEH (1995): "Emery's Elements of Medical Genetics," 9th Ed. New York: Churchill Livingstone.

Thompson MW, McInnes RR, Willard HF (1991): "Thompson & Thompson: Genetics in Medicine," 5th Ed. Philadelphia: W.B. Saunders Company.