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Genetics in medical school curriculum: A look at the University of Rochester School of Medicine and Dentistry

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Abstract: Genetics is assuming an increasingly important role in medicine. As a result, the teaching of genetics should also be increased proportionally to ensure that future physicians will be able to take advantage of the new genetic technology, and to understand the associated ethical, legal and social issues. At the University of Rochester School of Medicine and Dentistry, we have been able to incorporate genetic education into a four-year medical curriculum in a fully integrated fashion. This model may serve as a template for other medical curriculum still in development.

Key words: Genetics education, Medical education, Curriculum reform
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INTRODUCTION

The field of genetics has seen remarkable advancement in the past 50 years, from the discovery of DNA in 1953 to the completion of the Human Genome Project in 2003. This amazing progress has enriched the medical field greatly, and necessitated that all medical providers are fluent in the basic language and applications of genetics. For physicians to become adequately versed and remain current in human genetics, it is necessary to begin teaching the discipline in undergraduate medical education. Due to the remarkable pace of growth in genetics, this learning must continue on throughout the additional training years in addition to the years spent as a practicing physician. In this article, we will explore the undergraduate medical school curriculum of genetics utilizing the University of Rochester School of Medicine and Dentistry (URSMD) as a model.

In 2001, the American Society of Human Genetics (ASHG) and the Association for Professors in Human and Medical Genetics (APHMG) issued the "Medical School Core Curriculum in Genetics." This document outlined the necessary knowledge, skills and behaviors that all medical students will require in their future practice as physicians (http://genetics.faseb.org/genetics/aphmg/med-sch-guide.htm). The guidelines propose four manners to incorporate the teaching of genetics into the curriculum as follows:

(1) Medical genetics provides a unique perspective on function of the human body in health and disease; it is both a clinical specialty and a basic science. Medical genetics teaching must span the entire undergraduate medical school curriculum and continue into the postgraduate years.

(2) Medical genetics must be explicitly included in the curriculum. Although some aspects of medical genetics overlap with and may be taught by other disciplines, specific learning objectives in medical genetics need to be established.

(3) A well-qualified medical genetics specialist (or small committee of medical geneticists) should be given the authority and responsibility for implementing the genetics curriculum at each medical school. This responsibility should extend throughout the undergraduate medical curriculum and include involvement in all courses that deal with genetic principles or disorders.

(4) Medical genetics can be taught effectively by a variety of methods and in various formats. Problem-based learning (PBL) is particularly well suited to medical genetics because it involves integration of skills and knowledge from many fields. Genetics can also be taught in various clinical contexts and at different points in clinical training, depending on the particular circumstances at each school. Specific clinical examples are important, but the focus of the curriculum must be on medical genetic principles illustrated by the examples.

However, these are given as suggestions and thus, leave the specific implementation strategies up to the interpretation of the individual medical schools.

DOUBLE HELIX CURRICULUM AT UNIVERSITY OF ROCHESTER SCHOOL OF MEDICINE AND DENTISTRY

At URSMD, the entire medical school curriculum underwent a significant reform in 1999. The traditional two-plus-two, largely lecture-based medical curriculum (two years of didactic basic science followed by two years of clinical immersion) of the Flexner era gave way to an appreciation of the importance of training physicians who are adaptable, life-long learners who possess the skills and attitudes necessary to remain competent, and humanistic physicians in an age characterized by a tremendous explosion of new medical knowledge (Ludmerer, 1999). This change refocused the medical education on helping students "learn how to learn." The curriculum reform resulted in the Double Helix Curriculum (DHC), named because it consists of "intertwining strands" of basic and clinical science education throughout the four-year curriculum. The DHC is focused on developing life-long learning in students in addition to biopsychosocial medicine, which has been a hallmark of the education of medical students at University of Rochester since the 1940's. This curricular and focal change was particularly pertinent for a field such as medical genetics, due to the rapidity of progress being constantly made in such areas.

The DHC captures the integrated strands of basic science and clinical medicine interwoven throughout a four-year curriculum. Although of faculty design, the DHC is a "hybrid PBL" curriculum that is centrally administered (Reynolds *et al*., 1995). Every course is interdisciplinary: basic sciences are integrated with one another and basic and clinical sciences are woven together as the strands of the DHC (Fig.1, http://www.urmc.rochester.edu/smd/education/medical/curriculum.cfm). The clinical strand occupies approximately 30% of curricular time in Years 1 and 2, and 80% in Years 3 and 4, with the basic science strand representing the converse.

Highlights of the DHC include:

(1) Basic science and clinical education integration across all four years: four years of basic science and four years of clinical science.

(2) History and physical examinations skills taught in the first semester (Introduction to Clinical Medicine) followed by 18 months of Ambulatory Care Clerkship (ACE).

(3) Year 1 courses also include Managing Medical Information (statistics, epidemiology, information technology skills), Human Structure and Function (anatomy, physiology, histology), Molecules-to-Cells (biochemistry, genetics, cell biology), Host Defense (microbiology, immunology, basic pathology, dermatology), and Medical Humanities Seminar.

(4) Year 2 courses also include Mind, Brain and Behavior (neuroscience and behavioral science), Pharmacology, Disease Processes and Therapeutics (systems pathophysiology), Medical Humanities Seminar, and Case Seminars (assorted basic science topics).

(5) Year 3 Core Clinical Clerkships: Medicine, Surgery, Women's and Children's Health (obstetrics/gynecology and pediatrics), and Mind, Brain, Behavior (neurology and psychiatry). At the end of the latter three Core Clerkships are three Advanced Basic Science blocks. The remainder of the curriculum is filled with Electives.

(6) Year 4 curriculum consists of Emergency Medicine, Community Health Improvement Clerkship, Elective and Clinical Sub-Internships. It also consists of the month-long Process of Discovery course which teaches students how to design research studies, formulate grant applications and give meeting presentations. The year ends in the Successful Interning course which provides additional clinical skills for students to prepare them for clinical internship and residency after graduation.

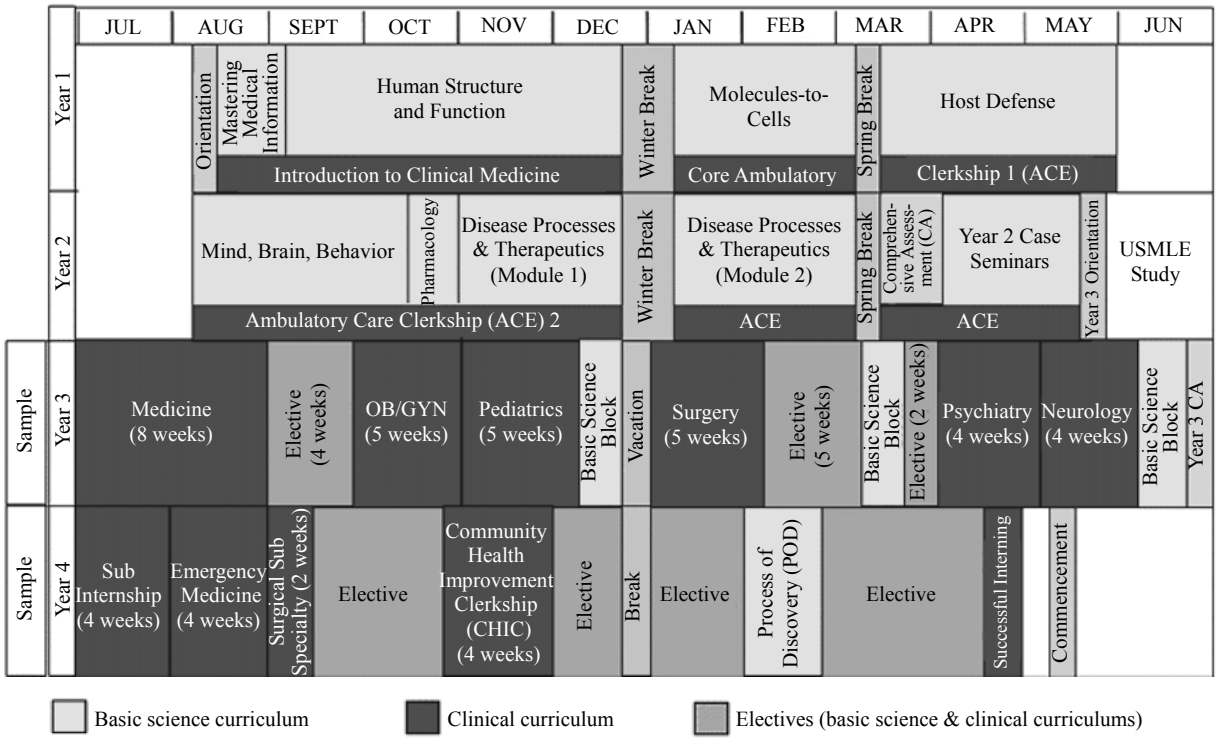

Fig.1 Map of the Double Helix Curriculum at the University of Rochester School of Medicine and Dentistry
OB/GYN: Obstetrics/gynecology; USMLE: United States Medical Licensing Examination™

(7) Mix of weekly PBL sessions and lectures, labs, and small-group conferences for all basic science courses.

(8) Comprehensive Assessments at the end of Years 2 and 3, resulting in the development of an Individualized Learning Plan for each student.

(9) Emphasis on six crosscutting themes: Aging, Diversity, Ethics and Law, Health Systems, Nutrition, and Prevention.

MEDICAL GENETICS CURRICULUM WITHIN THE DHC

Thus the genetics curriculum at URSMD was constructed with a few goals in mind. Firstly, it should adhere to the ASHG/APHMG guidelines. Secondly, it is consistent with curriculum integration goals of the DHC, and to comply with its adult learning mission. Thirdly, it should provide students with enough basic genetics knowledge to "service" subsequent courses in the curriculum. Lastly, it should enhance the teaching of the six cross-cutting themes in the DHC.

In the DHC, medical genetics is not taught as a single course, but can be considered a "motif" that is part of many different courses and themes. While most of what is considered traditional medical genetics is taught in the first year course Molecules-to-Cells, which combines biochemistry and genetics, genetic content permeates throughout the entire four years in many courses. The genetic curriculum content for each course in the four-year curriculum is shown in Table 1.

Topics essential to understanding genetics are first encountered by beginning first-year students in the primer course of their medical education, Mastering Medical Information. In this epidemiology, biostatistics, evidence-based medicine, and clinical trial design course, the students are taught the statistical principles that form the foundation for understanding population genetics and genetic screening in addition to the introduction of common complex disorders. Similarly, in Human Structure and Function, an Anatomy-Physiology course in Year 1, embryology is first presented. These topics are specifically followed up during the Molecules-to-Cells course: population and family genetics data are

Table 1 Medical genetics curriculum content in the Double Helix Curriculum, University of Rochester School of Medicine and Dentistry

Year	Course	Content
Year 1	Introduction to Clinical Medicine	Taking a family history
Year 1	Managing Medical Information	Statistics; epidemiology
Year 1	Human Structure and Function	Embryology
Year 1	Molecules-to-Cells	Molecular genetics: (1) Human genome structure, genome evolution, mutation; (2) Human gene structure and control of gene expression; (3) DNA replication and repair, recombination; (4) Chromatin structure, gene regulation and transcription; (5) RNA processing; (6) Translation and special RNAs; (7) Post-translation modification (folding and glycosylation); (8) Proteins structure and disorders (collagen, fibrillin, hemoglobin) Population genetics and genetic anthropology: (1) Hardy-Weinberg law; (2) Inbreeding and consanguinity; (3) Genetic admixture, migration, gene flow; (4) Mutation and mutation rate; (5) Evolution of human populations; (6) Genetics of race Medical genetics: (1) Modification of Mendelian pattern of inheritance; (2) Clinical cytogenetics and chromosomal disorders; (3) Mitochondrial genetics; (4) Molecular diagnostics; (5) DNA cloning; (6) Newborn screening; (7) Prenatal diagnosis; (8) Genetic ramifications of fertility treatment; (9) Genetic counseling and Bayes theorem Developmental genetics: (1) Cell cycle and growth control; (2) Basic developmental genetics (cell fate, body plan, patterning, cell movement and migration); (3) Cell signaling and genes involved in developmental pathways (receptor tyrosine kinases, FGF, TGF, Notch, Sonic hedgehog, Wnt); (4) Teratology and dysmorphogenesis; (5) Genome imprinting; (6) Stem cell biology and medical applications; (7) Transgenic mice Cancer genetics: (1) Oncogenes/proto-oncogenes; (2) Tumor suppressor genes and other cancer critical genes; (3) Cancer cytogenetics; (4) Apoptosis; (5) Tumor angiogenesis, hypoxia and therapeutic resistance; (6) Development, cancer and aging "Omics": (1) Mapping of Mendelian traits; (2) Human genome sequencing and the Human Genomic Projects; (3) Mapping common complex traits and genetic association; (4) Linkage disequilibrium (LD), LD mapping and the HapMap; (5) Nutrigenomics; (6) Pharmacogenetics and pharmacogenomics; (7) Proteomics; (8) Network biology Patient presentations: (1) Down syndrome; (2) Ehlers-Danlos syndrome; (3) Fragile X syndrome; (4) Sickle cell disease; (5) Hereditary non-polyposis colon cancer (HNPCC); (6) Galactosemia; (7) Long-chain hydroxyacyl dehydrogenase (LCHAD) deficiency; (8) Type 1 diabetes; (9) Phenylketonuria; (10) Lipoprotein lipase deficiency; (11) Hurler-Scheie disease; (12) Crouzon syndrome; (13) Ataxia telangiectasia 8 PBL cases: (1) Hereditary non-polyposis colon cancer; (2) Glycogen storage disease type 1b; (3) Glutaric aciduria type 2 (multiple dehydrogenase deficiency); (4) Ornithine transcarbamylase deficiency; (5) Apo B100 deficiency (familial hypercholesterolemia variant); (6) Smith-Lemli-Opitz syndrome; (7) Neuroblastoma; (8) Diabetes mellitus type 1

(continued in the next page)

Year	Course	Content
Year 1	Host Defense	Immunogenetics; microbial genetics
Year 2	Mind, Brain and Behavior	Neurogenetics disorders
Year 2	Disease Processes and Therapeutics	Pathogenesis of various genetics disorders; prenatal diagnosis and artificial reproductive technology
Year 2	Year 2 Case Seminar	Practical clinical cytogenetics; practical molecular diagnostics
Year 3	Core Clinical Clerkships	Opportunistic patient encounters
Year 3	Women's and Children's Health Clerkship Advanced Basic Science Block	Integration of clinical issues and basic science: (1) Genetics of infertility; (2) Sex determination; (3) Evolution of sex; (4) Genomic imprinting and epigenetics; (5) Teratology; (6) Hands-on syndrome diagnosis exercise; (7) Genetics of developmental pathways and testing; (8) Neurodevelopmental disorders; (9) Biology-Ecology-Culture paper
Year 3	Elective Clinical Genetics Rotation	Clinical genetics
Year 3	Elective Craniofacial Team Care Elective	Craniofacial genetics
Year 3	Comprehensive Assessment	Case of breast cancer
Year 4	Electives	Opportunistic patient encounters
Year 4	Elective Clinical Genetics Rotation	Clinical genetics
Year 4	Process of Discovery	Students work in small groups to write a grant proposal for research to help patient with one of two clinical problems (e.g. osteoporosis, cardiomyopathy, neuroprotection), with many research designs involving genetics approaches; co-evolution of genes and culture

analyzed using the chi-squared method; Bayesian statistics are used to solve genetic counseling cases, and normal distribution is used to illustrate common complex genetics. Furthermore, clinical genetics skills such as obtaining family history are addressed in Introduction to Clinical Medicine and Ambulatory Care Clerkship courses. This skill is specifically reinforced during the Comprehensive Assessment at the end of Year 2.

The Molecules-to-Cells course in Year 1 is a 10-week course that meets for 4 h/d. It consists of 7 major theme blocks: biochemical basics, intermediary metabolism, molecular genetics and cell structure, cell growth control and cancer, nutrition, selected pharmacology topics and medical genetics. The pairing of genetics and biochemistry is synergistic in the medical school setting. This distinctive combination allows for a more thorough and in-depth understanding of genetics in the backdrop of molecular cellular biology. For example, all medical students learn about phenylkentonuria (PKU) in regards to its autosomal recessive pattern of inheritance and the carrier frequency in various populations. However, when this information is combined with the intricacies of various enzymatic defects that cause PKU, and how patients with this disorder are managed from the nutritional, emotional and educational standpoints, a more holistic understanding of the condition can be appreciated. Another illustration of how the combination of genetics and biochemistry is advantageous can be seen in conditions of developmental delay, of which only hypotheses as to the etiologies are proposed. In many of these cases, intracellular signaling pathways are thought to play a role in the conditions, and thus, the conditions can be better comprehended with an appreciation of both biochemistry and genetics.

Another distinctive characteristic of the Molecules-to-Cells course is the emphasis placed on patients. Many times the students are exposed to more than just a lecture on a particular syndrome; they are also afforded the opportunity to meet patients who are living with these conditions. For instance, an hour lecture is devoted to Crouzon syndrome after which there is an hour of meeting with a young patient who has Crouzon and his or her parents. This exposure not only aids in the consolidation of knowledge, but reiterates the biopyschosocial nature of patients and their conditions. The students become aware of more than a craniofacial developmental disorder of autosomal dominant inheritance; they become intimately aware of the quotidian life of these patients and their

parents and how their existence is similar to and different from those without Crouzon syndrome.

The integration of genetics into Years 3 and 4 has proven to be more challenging. Currently, in the 3rd year, genetics is encountered in the Advanced Basic Science Blocks, a forum which permits a back-to-basics approach that focuses on developmental biology. Students are given the clinical data, including patient photographs, to attempt to diagnose a dysmorphic syndrome. They are asked to provide a presentation to explain their reasoning, as well as the gene(s) and developmental pathway that is deranged in these patients. In addition, the 3rd year students are being asked to participate in a new assignment, in which they identify a patient from one of their core clerkships, and probe into the biological, ecological, and cultural factors that contribute to the disease that the patient is enduring. This approach of looking at a patient from biological, ecological and cultural viewpoints, provides a framework for students to meaningfully examine a patient in a biopyschosocial context. As a result, students are able to view the patient's illness as more than a biological defect and are thus, able to appreciate other factors that contribute to the patient's disorder. It also addresses attitudes and skills that a student must possess and practice in order to gather this information. In Year 4, genetics is once again seen in the Process of Discovery course; even though the genetics topics are more diffusely distributed, they are intertwined into the structure of the class.

As mentioned before, six themes are stressed in the DHC, with Diversity being one such topic. Through the theme of Diversity, genetics is smoothly integrated throughout the four years of medical education at URSMD. Under the Diversity theme, students and faculty are educated to become critically aware of concepts of race, class, gender, ethnicity, sexual orientation, and disability as social rather than solely biological constructs. As one can see, genetics provides a perfect vantage point to explore issues of human diversity and this is accomplished throughout the four years via PBLs, didactic sessions, readings, and small group and individual exercises.

CONCLUSION

At URSMD, we have created a four-year genetic curriculum that adheres to ASHG/APHMG recommendations and at the same time respects institutional educational priorities. From this perspective, the diverse forum where genetics is taught at URSMD serves to reiterate genetics learning, as well as providing a wide array of formats in which genetics is taught, including lectures, small group discussions, large group patient demonstrations and PBLs. We find that this integrative and encompassing manner in which genetics is taught facilitates not only the necessary fundamentals of genetics but more importantly emphasizes life-long learning strategies. This is of critical importance as the rate of progress in the field of genetics is accelerating at a feverish pace.

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