

Developing a Blueprint for Primary Care Physician Education in Genomic Medicine
June 8-9, 2009
Meeting Report

I. Represented organizations

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)
Centers for Disease Control (CDC)
Health Resources and Services Administration (HRSA)
National Cancer Institute (NCI)
National Heart, Lung and Blood Institute (NHLBI)
National Human Genome Research Institute (NHGRI)
National Newborn Screening and Genetics Resource Center (NNSGRC)
NIH Office of Rare Diseases Research (ORDR)
Advisory Committee on Heritable Diseases in Newborns and Children (ACHDNC)
Albert Einstein College of Medicine
American Academy of Family Physicians (AAFP)
American Academy of Pediatrics (AAP)
American Association of Colleges of Osteopathic Medicine (AACOM)
American College of Medical Genetics (ACMG)
American College of Obstetrics and Gynecology (ACOG)
American College of Physicians (ACP)
American College of Preventive Medicine (ACPM)
American Medical Association (AMA)
American Osteopathic Association (AOA)
Association of American Medical Colleges (AAMC)
National Coalition for Health Professional Education in Genetics (NCHPEG)
National Institute of Child Health and Human Development (NICHD)
National Society of Genetic Counselors (NSGC)
Physician Assistant Education Association (PAEA)
Secretary's Advisory Committee on Genetics, Health and Society (SACGHS)
Society of General Internal Medicine (SGIM)
Society of Teachers of Family Medicine

II. Introduction

The “Developing a Blueprint for Primary Care Physician Education in Genomic Medicine” meeting was held on June 8-9, 2009 at the National Institutes of Health (NIH). The meeting was co-sponsored by: the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), the Centers for Disease Control (CDC), the Health Resources and Services Administration (HRSA), the National Cancer Institute (NCI), the National Heart, Lung and Blood Institute (NHLBI), the National Human Genome Research Institute (NHGRI), the National Newborn Screening and Genetics Resource Center (NNSGRC), and the NIH Office of Rare Diseases Research (ORDR).

The meeting was convened to provide an opportunity for primary care physician leadership as well as leadership from the genetics community to discuss implications of genetics/genomics discoveries for their practice and education needs. Primary care is

undergoing significant upheaval, including a potential reorganization to focus on the medical home. Certainly, the medical home of the 21st century will need to deal with substantial advances in our understanding of the genetic contribution to health and disease. Improved health outcomes from genetic discovery will only be realized if the healthcare community is aware of and can implement the opportunities brought about by genome research.

The stated goals of the meeting were as follows:

- 1) To define the current landscape of primary care education in genetics and genomics from the perspective of genetics, primary care, and academic medical communities.
- 2) To identify core educational needs in genetics and genomics as defined by primary care communities.
- 3) To identify opportunities and barriers that face efforts to enhance primary care provider genetic/genomic literacy.
- 4) To propose concrete strategies to take advantage of existing opportunities for genetics/genomics education at various stages of primary care physician education.
- 5) To plan next steps.

The proposed work product from the meeting was a five-year blueprint for improving primary care clinician literacy in genetics and genomics that is informed by the realities facing primary care. Here, we summarize the recommendations for primary care physician education in genomic medicine at all career levels that were generated at the meeting.

II. Meeting content

The one and a half day meeting consisted of a mix of presentations and small group sessions. The presentations on day one consisted of a brief introduction to the advances in genomics relevant to primary care over the last five years, as well as the looming issue of direct to consumer marketing of genetic testing. During the noon sessions representatives of both nursing and physician assistant communities presented an overview of the genomics educational activities in their respective disciplines. Group discussion sessions covered current and past genomics education activities directed toward primary care physician education in genomics. The first day breakout sessions covered a discussion of the genomics educational needs of various types of primary care providers, as well as the facilitators and barriers of genomics education for primary care physicians.

Day two included presentations that demonstrated a number of genomics educational activities developed for health care providers. The second day breakout session consisted of discussions of concrete steps that could be undertaken to advance primary care provider genomics education for all levels of learners (medical school, residency, and the practicing provider). Day two wrapped up with a session that covered educational issues specific to providers of maternal child health services.

Specifically breakout sessions addressed the following topics:

Day one –

Priority Setting

- A. What are the core genetic/genomic educational needs for all primary care specialties in the next five years?
- B. What are the core genetic/genomic educational needs unique to each primary care specialty in the next five years?
- C. What are the barriers and potential facilitators of primary care education in genetics?

Day two –

Five Concrete Steps for the Next Five Years

- A. Medical student education
- B. Resident education
- C. Practicing clinicians

III. Outcomes from breakout sessions:

Day One- Priority Setting

A. Suggested educational needs for all primary care physicians

In order to respond to the continuously expanding understanding of the contribution of genetics/genomics to health and disease, primary care physicians in all specialties need education to develop skills/knowledge in the following core areas:

- Fundamentals of medical genetics (e.g., patterns of inheritance, understanding genetic testing)
- How to take, update, and use a family history
- Assessment, communication, and management of risk relevant to family history and genetic testing where appropriate, including
 - Clinical epidemiology
 - Biostatistics
 - Population/individual risk, racial/ethnic differences
- The importance of gene-environment interactions and the role of behavior
- Making appropriate patient referrals for genomic issues
- Appropriate interpretation and translation of physician-ordered and direct-to-consumer (DTC) genetic tests
- Pharmacogenetics
- Ethical, legal, and social implications (ELSI) of genetic/genomic testing

B. Suggested primary care specialty-specific needs:

Though the groups acknowledged that there was significant overlap in the genomics educational needs of the primary care specialties it was recognized that some groups required additional, or at a minimum more in-depth content covering specific aspects of care. In addition to the core genetics/genomics needs of all primary care physicians, the primary care specialties have specific educational needs:

i. Obstetrics/Gynecology (OB-GYN)

- **Pre-conception care**
Pre-conception care is a part of obstetrics/gynecology, internal medicine, and pediatrics. It should include a genetic/genomic component, which in obstetrics/gynecology may consist of examining the prospective parents' family histories, performing genetic tests, and referring patients to other specialists or genetic counselors.
- **Prenatal diagnostic testing options**
Physicians should be aware of the available prenatal diagnostic tests and have an understanding of the appropriate conditions for ordering the tests.
- **Newborn screening**
As with prenatal diagnostic testing, physicians should be aware of the available newborn screening tests, including screening for late-onset diseases. Newborn screening is an element of the transition between OB-GYN and pediatrics, as the mother's and newborn's physicians need to be made aware of the results of the tests.
- **Women's health**
Like pre-conception care, women's health care can begin in pediatrics and is a part of both internal medicine and obstetrics/gynecology. The genetic/genomic component of women's health care includes examining family history and performing genetic tests (e.g., breast cancer)

ii. Pediatrics

- **Newborn screening**
- **Effect of genetics on growth and development**
- **Genetics/genomics of mental health**
- **Pre-conception care**
Although it is debatable whether pre-conception care is part of pediatrics, this care could begin as early as age 9 and is an element of the eventual transition of care from pediatrics to internal medicine and/or OB-GYN

iii. Internal Medicine

- **Genomics content regarding women's health (e.g., hereditary breast and ovarian cancer syndrome, endometrial cancer and Lynch syndrome)**
- **Genetics/genomics of mental health**
- **Pre-conception care**
Genetic/genomic pre-conception care in internal medicine may include making patients (female and male) aware of potential genetic/genomic considerations if they should decide to have children.
- **Interfacing with sub-specialties regarding genetic testing.**
The bulk of genetic testing is performed within the subspecialties of internal medicine (e.g., gastroenterology, rheumatology, oncology). Internists need the educational background to enable them to interface effectively with subspecialists regarding genetic tests and patient care.

iv. Geriatrics

Geriatrics was considered distinct enough by the groups to warrant special consideration.

- Pharmacogenetics and polypharmacy
Pharmacogenetics and polypharmacy have particular importance in geriatrics because patients are more likely to be taking at least one medication.
- Family history (note this is core to all specialties but often not considered in geriatrics)
The ability to take a family history is an important skill for all primary care, and there is an opportunity in geriatrics to develop a more complete family history.
- Genetics/genomics of aging
- Older patients are the least likely to benefit from genetic testing
- Genetic testing in geriatric patients may be difficult to justify and have costs covered.

v. Family Medicine

Family medicine has all of the same specialty-specific needs as the other primary care specialties.

- Family physicians are more likely to practice in rural areas, where access to resources, like genetic counselors, may be problematic. They may require more in-depth skills and the ability to more effectively access genomic information/resources relevant to patient care.

C. Barriers and facilitators

i. Barriers

Barriers to primary care education in genetics/genomics are substantial. Perhaps the most problematic barrier is a persistent misperception that genetics/genomics is not relevant to primary care practice. This is in part because of a lack of provider knowledge, and partially because there is a paucity of research demonstrating the effectiveness of genomic applications in primary care environments. There is a lack of knowledgeable and supportive faculty and leadership during the training of primary care providers which makes it difficult to quickly improve primary care training programs.

The predominance of the acute care model (vs. chronic or preventative care models) in many primary care settings puts tremendous time pressures on primary care providers. This model of care fails to reward the adoption of potentially complex and time consuming genetic/genomic applications to enhance care.

Attendees also recognized a lack of financial resources to devote to genomics educational material development and implementation within their organizations.

ii. Facilitators

Evidence of the effectiveness of the incorporation of genetics/genomics into primary care could be a strong facilitator of primary care education in and incorporation of genetics/genomics. Evidence of effectiveness may convince primary care physicians to pursue genetics/genomics education and incorporate the field into their practices, overcoming the practical barriers described above. However experience suggests that evidence of benefit of any health application is by itself insufficient to ensure widespread adoption.

Other resources which could facilitate primary care education in genetics/genomics are:

- Knowledgeable faculty and primary care leadership
- A shift to a patient-centered medical home model in which preventive services are emphasized and rewarded
- Family history and genomic clinical decisions support tool integration into EHR systems
- Financial or other incentives for education, proficiency, and practical application of genetics/genomics in primary care

iii. Needs

The attendees recognized that effective genomics education of primary care physicians will require that certain infrastructure/organizational needs be met. Those discussed, in no defined order included:

- Faculty at all levels trained in genetics/genomics
- Increased genetics content in boards and licensing exams
- Effective communication of new information/evidence to medical students, residents, and physicians
- Better evidence supporting the value of genomic applications in practice
- Development of strategies for the translation of knowledge to practice
- Increased focus of genomic technologies on primary prevention
- Increased integration of genetic/genomic competencies across all phases of training
- Updated, evidence-based guidelines for use of genomic applications
- Reimbursement for the time required to provide genomic services including family history use and counseling
- Improved infrastructure for transitions in care of the course of a patient's lifetime
- Clinical decision support tools to support busy clinicians
- Electronic patient-oriented tools

Day two – Five Concrete Steps for the Next Five Years

A. Medical Students

Medical genetic/genomic education begins in medical school and should form a strong base for later, specialty-specific training. The meeting participants made the following recommendations for the education of medical students in genomic medicine:

- Evaluation and oversight of the curriculum in genetics and genomics
 - Participants recommended that curriculum committees evaluate medical school courses for content as it relates to genetics and genomics. This content should include:
 - a. Fundamentals of genetics/genomics (core knowledge)
 - b. Medical/clinical genetics/genomics and their relevance to patient care
 - c. Screening, testing, epidemiology, risk, and biostatistics (clinical epidemiology)
- Faculty development (didactic and clinical)
 - Participants recommended that medical schools increase their number of educators capable of presenting genetics/genomics materials, both in the didactic and clinical years. In addition, faculty who do not feel sufficiently prepared to incorporate genetics/genomics in their instruction should be identified, and resources should be provided for the identified faculty members.
- Integration of genetics/genomics throughout the 4-year training period
 - Participants agreed that genetics/genomics instruction should occur during all four years of medical school education. With respect to didactic instruction, there are multiple potential methods of providing genetics/genomics instruction: independent curriculum, integration in existing courses, and problem-based learning. Participants also specifically recommended that genetics/genomics be integrated in clinical year instruction, through clerkships, electives, and/or other avenues. The curriculum chair could assist in the assessment and assurance of integration of genetics/genomics throughout all courses.
- Adaptation of the Liaison Committee on Medical Education (LCME)
 - Changing genetics/genomics education of medical school students will be facilitated by support from the LCME and changes in the content and scoring of the United States Medical Licensing Examination (USMLE) Steps 2 and 3 with respect to genetic/genomic content.
- Ongoing evaluation and modification of genomics educational program outcomes as knowledge of genetics/genomics evolves
 - As genetics/genomics education in medical schools and the genetics/genomics field itself evolve, medical schools will need methods for evaluating their success in educating students and modifying their curricula in response to those outcomes. Changes in the LCME and USMLE as described above will be a part of this process.

B. Residents

Education of residents in genetics/genomics has cross-specialty and specialty-specific components. The meeting participants made the following recommendations for the education of residents in genomic medicine:

- Involve the groups that govern resident education: Accreditation Council for Graduate Medical Education (ACGME), medical boards, and residency review committees (RRC).
 - ACGME
Participants suggested approaching the ACGME with united (across specialties) suggestions for updating the genetics/genomics education of residents.
 - Medical Boards
Participants suggested asking them to include genetics/genomics questions for certification and maintenance of certification. It was acknowledged that there may be resistance to changing the requirements for board certification due to the evolving nature of the field.
 - RRC
Participants suggested identifying and cultivating genetics/genomics education “champions” on RRCs. There was consensus that in no way should genomics be introduced to the resident training process as “added requirements,” but rather mapped to subject matter already being delivered.
- In order to convince residents and educators of the importance of genetics/genomics education, it will be necessary to demonstrate its relevance to their specialties.
- The development of a genetic toolkit for educators, like the Genetics in Primary Care (GPC) program, would support resident training in genetics/genomics. This tool kit could include standardized patient scenarios that illustrate relevance of clinical genomics to all specialties. A genetics/genomics “speaker’s list” could be developed and made available as a resource for institutions without expertise in specific content areas.
- Consider developing advanced genetics/genomics training programs for primary care residents
 - combined training with full board certification, such as current programs for pediatrics and medical genetics.
 - a one-year certification program similar to existing fellowship opportunities
 - genomics short-courses offering certificate of completion

- Board certification could require the demonstration of a level of proficiency in genetics/genomics.

C. Practicing Physicians

Practicing physicians are a more challenging audience to reach than medical students or residents. The meeting participants made the following recommendations for the general education of practicing physicians in genomic medicine:

- Assess the primary care professional group's approaches and activities regarding genomics education.
- Create evidence-based recommendations and guidelines that are agreed upon across primary care disciplines. This activity will require communication across the professional groups, and could include interdisciplinary consensus group meetings, development of a collaborative repository of information, and a system for disseminating relevant new genomics information across disciplines.
- CME certification should include a genetics/genomics component
- Develop and make available a family history tool with electronic medical record (EMR) integration and clinical decision support. Family history was acknowledged as a stepping stone for primary care genomics education, but time saving and systematic approaches to facilitate collection and interpretation must be developed.
- Create effective primary care friendly tools for clinicians and patients to support informed decision-making.
 - Evidence based, point of care tools embedded in EHRs, on the internet, and on PDAs
 - Accessible information (e.g., Gene Facts, Gene Clinics) which provide basic information on syndromes and common diagnoses
 - Accessible unbiased information regarding direct to consumer genetic testing
 - Information for patients to facilitate shared decision making
 - Create tools to emphasize that genomics has a strong influence on health care across the lifespan.

IV. CONCLUSIONS AND NEXT STEPS

At the “Developing a Blueprint for Primary Care Physician Education in Genomic Medicine” meeting held on June 8-9, 2009, representatives from various government organizations, physicians' groups, and genomics/genetics groups met to discuss the education of primary care physicians in genetics/genomics. The meeting participants identified the core genetics/genomics education needs for primary care physicians and specialty-specific needs. Attendees identified barriers as well as facilitators of primary care education in genetics/genomics. Finally, they proposed a variety of ways to enhance

genetics/genomics education for physicians throughout the course of their training. Overall the meeting was a successful first step towards promoting primary care physician genetics/genomics education.

Proposed next steps:

- Development of organization-specific strategies including three- and five-year goals for genetics/genomics education over the next three to six months
- Reconvening this group in six to eight months to discuss the feasibility of the organization specific strategies, and to look for areas ripe for interdisciplinary collaboration
- Presenting the strategies to a wider audience of primary care education stakeholders at a meeting in 12 months
- Implementing developed strategies over next 12-36 months with annual update meetings involving relevant organizations.

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