Genetic and Genomic Literacy in Pediatric Primary Care

abstract

A colloquium on genetic literacy in pediatric primary care sponsored by the Health Resources and Services Administration Maternal and Child Health Bureau was held at the American Academy of Pediatrics headquarters on October 2–3, 2012. The overarching goal of the colloquium was to provide context for delivery of genetics-related services in day-to-day pediatric primary care practice, encompassing 3 dimensions of medicine: prevention, diagnosis, and management. Participants considered the whole spectrum of disease, from rare disorders to common disorders, the genetics-related components of which are often overlooked. Specific topics included family history, genomics, genetic literacy and competency, epigenetics, and a focused view of primary care and genetics. A consensus statement was developed to provide recommendations for integration of genetics into pediatric primary care. *Pediatrics* 2013;132:S198–S202

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KEY WORDS

family history, genomics, genetics, genetic literacy, genetic competency, epigenetics, pediatric primary care, public health

ABBREVIATIONS

AAP—American Academy of Pediatrics
GPCI—Genetics in Primary Care Institute
PCP—primary care provider

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EXECUTIVE SUMMARY

Without question, man's knowledge of man is undergoing the greatest revolution since Leonardo.... In many ways, personalized medicine is already here.

—Dr. Francis Collins

Director of the Human Genome Project and the US National Institutes of Health

The completion of the Human Genome Project in 2003 was one of humankind's greatest scientific accomplishments. For the first time, it is possible to read nature's complete genetic blueprint. As genomic science has advanced, the focus of genetic and genomic medicine has shifted from single-gene, rare conditions to common chronic conditions. Recent advances in the fields of genetics, genomics, and epigenetics have increased our understanding of the interplay between genetic and environmental factors as determinants of health. On average, 5 new genetic tests are developed each month,[1] adding to the 1300 already available,[2] and new treatments continue to emerge for genetic conditions for which there were previously none. Today's medical practitioners must engage in genome-wide thinking and the practice of personalized medicine. Just as advances in the prevention, diagnosis, and treatment of infectious diseases in the 20th century led to dramatic advances in medical care, so too does genomic medicine promise to provide similar enhancements to medical care and population-based health in the 21st century.

The American Academy of Pediatrics (AAP) has identified a need to ensure that personalized medicine can provide care that is tailored to the unique genetic traits of the individual. The Genetics in Primary Care Institute (GPCI), a 3-year cooperative agreement between the Health Resources and Services Administration Maternal and Child Health Bureau and the AAP, has been established to meet this need in a patient's medical home where care is delivered by a primary care provider (PCP). PCPs are the health care professionals to provide comprehensive health care to patients and their families, fulfilling the primary health care needs unless specialty care is also needed from a specialist.

The overall goal of the GPCI (www.geneticsinprimarycare.org) is to increase PCPs' genetic literacy within the following priority areas:

- Understanding of basic genetic principles
- Collecting and interpreting family history
- Engaging patients and their families as active partners
- Ordering, interpreting, and acting on genetic tests
- Establishing partnerships with genetics experts within a community
- Enhancing education about emerging technologies and tests
- Navigating ethical considerations regarding genetics-related issues
- Integrating the provision of genetic medicine into health information technology

The GPCI sought to engage a group of thought leaders and experts to review the salient issues and to formulate recommendations to improve the genetic literacy of pediatric PCPs. This group gathered at the Genetic Literacy in Pediatric Primary Care Colloquium, sponsored by the Health Resources and Services Administration Maternal and Child Health Bureau and held on October 2–3, 2012, at the AAP headquarters in Elk Grove Village, Illinois.

The theme of the colloquium was to provide context for genetics in day-to-day primary care practice. Instead of encouraging providers to learn endless facts about genetics and genomics, which quickly become outdated, emphasis was placed on improving PCPs' genome-wide thinking and then encouraging them to access genetic and genomic information when needed. The colloquium addressed the key issues regarding genetic and genomic medicine in primary care, illustrating the roles of pediatricians and other PCPs in the emerging world of genomic medicine while encompassing 3 dimensions of medical practice (eg, prevention, diagnosis, and management) and considering the whole spectrum of disease type, from rare to common disorders. To establish common ground, the authors came to a consensus regarding the definitions of 3 key concepts: genetics, genomics, and epigenetics:

- **Genetics** describes the study of genes and their role in inheritance—the way certain traits or conditions are passed down from one generation to another.
- **Genomics** describes the study of all of a person's genetic material, the interactions among the components of that genetic material, the interactions of that material with the environment, and the resulting phenotypic changes. Genetics uses information from 1 or 2 genes to explain a disease state, whereas genomics examines all of the genetic information to determine biological factors that might predispose an individual to disease. The terms "genetics" and "genomics" represent a continuum of genetic analysis.
- **Epigenetics** refers to the study of functionally relevant chemical modifications to DNA and genomic proteins that do not involve a change in the DNA nucleotide sequence.

The 5 articles in this supplement arose as a natural result of 5 areas of focus in the colloquium (see below) and address the key issues regarding genetic and genomic medicine in primary care, illustrating the roles of pediatricians and other PCPs in the emerging world of genomic medicine.

The overall title of the colloquium, "Genetic Literacy in Primary Care," is well-served by these focus areas, emphasizing that the broad topic of genetic literacy in primary care will require a multifaceted approach moving forward. The manuscripts address 5 major facets of care with which PCPs should be familiar: (1) family history, (2) genomics, (3) epigenetics, (4) genetic literacy and competency, and (5) primary care and genetics. Within each article, the integration of genetics and genomics into pediatrics is discussed in regard to each of the focus areas and within the context of diagnosis, treatment, and prevention (where pertinent). A brief summary of the manuscripts follows:

- The first article, "Family History in Primary Care Pediatrics," addresses the use of 1 of the most basic tools in the PCP's armamentarium, the family history. Although recognized for its vital information in patient care, the family history is still variably collected and used the primary care setting. This challenge is addressed, and alternatives are considered.
- The term "genetics" is the age-old term used to refer to genetic information. The more precise term, "genomics," has now emerged on the clinical scene, but many PCPs are likely baffled by it. The second article, "Whole Exome/Genome Sequencing and Genomics," provides the brief but necessary technical basis for understanding the concepts of genomics and then discusses the early stages of the integration of these technologies into primary care. Genomics and exome/genome sequencing will have a dramatic impact in the years ahead but in a way yet to be determined.
- Another confusing term that has entered the genetic literacy arena is epigenetics. Although this term as defined above might appear to be separate from genetics and genomics, it is a logical part of the whole of genetics/genomics and represents a specific burgeoning field exploring the integration of genetics/genomics and environmental influences. The third article, "Epigenetics and Primary Care," discusses the scientific bases for epigenetics and the implication that a significant component of common complex childhood diseases (such as obesity, asthma, and diabetes)

TABLE 1 Genetic Literacy in Primary Care Colloquium Consensus Statement

Recommendations for integration of genetics into primary care
1. Define how pediatric PCPs should use genetics and genomics in practice
   a. Recognize that many PCPs already use genetics and genomics in their practice; what is needed is evolutionary progress, not revolutionary change
   b. Approach primary care using the framework of a medical home model; genetics and genomics can augment and strengthen this model
   c. Emphasize the development of competencies in genetics and genomics, many of which can be mapped to competencies that have already been incorporated into training
2. Define, develop, and provide the tools and resources that are needed to integrate genetics and genomics into primary care
   a. Emphasize the relative values of targeted and comprehensive family histories and provide tools to facilitate collecting each
   b. Facilitate and encourage point-of-care use of relevant and credible genetic and genomic information resources
   c. Create point-of-care decision support tools for the use and interpretation of patients' genetic and genomic information
   d. Provide patient and family education and support tools that are culturally sensitive as well as literacy and language appropriate
   e. Facilitate access to appropriate family support and advocacy
3. Integrate genetics and genomics into primary care training at all levels
   a. Identify the fundamental concepts of genetics and genomics that are important to primary care practice
   b. Incorporate genetics and genomics into professional competencies
   c. Recognize that genetics and genomics educational efforts must span the entire educational continuum, from the preprofessional to the postgraduate level
4. Provide an evidence base for optimal integration of genetics and genomics into primary care
   a. Identify gaps in the evidence base regarding genetics and genomics in primary care
   b. Develop a research agenda to help fill these gaps
   c. Identify existing and needed infrastructure to facilitate the research agenda

TABLE 2 Evidence Gaps for Genetic Literacy in Primary Care[3–5]

1. Establishing genetic competency standards for training all primary care providers
   a. What are the essential skills each provider must master?
   b. How can these skills enhance existing linkages between primary care providers and their colleagues in subspecialty fields (including genetics counseling) and public health?
   c. How do these skills integrate with existing Accreditation Council for Graduate Medical Education competencies?
2. Supporting genetic literacy of patients and families in primary care
   a. What specific tools for discussing genetic and genomic information are most likely to improve communication in the primary care setting?
   b. What are the essential aspects of a patient's or caregiver's genetic literacy?
      i. What must be understood to satisfy informed consent before genetic testing?
      ii. How can test results (eg, risk of future illness, uncertainty) be most effectively communicated?
   c. How can genomic information be communicated clearly and with sensitivity to cultural differences (eg, language, literacy, ethnicity)?
3. Assessing clinical validity and utility of genetic and genomic tools in primary care
   a. What essential components of individual-level genetic information (eg, family history, genetic biomarkers, epigenetic profile) are most
      i. clearly associated with improved outcomes,
      ii. clearly associated with modifiable risk for future illness, and
      iii. critical at each life stage (the preconceptional period, the prenatal period, infancy, early childhood, adolescence, and early adulthood)?
   b. How can these tools best be integrated with the family-centered medical home model?
   c. Does the use of genomic information improve health outcomes, reduce unnecessary use of health care services, and/or reduce the costs of care?

are due to epigenetic factors that are still being investigated. The article also discusses how epigenetic factors might be directly relatable to disorders that PCPs deal with every day.

- The fourth article, "Genetic Literacy and Competency," specifically addressing the main title of the colloquium, recognizes the difficult work ahead as we try to translate ideas about genetics and genomics into specific action plans for medical education and physician training and maintenance of certification and presents a conceptual framework for moving forward.
- The final article, "Primary Care and Genetics and Genomics," reviews some of the historical issues regarding primary care and genetics and genomics and weaves its way through current times, providing a variety of resources to be considered. The key points presented emphasize how PCPs should be interfacing with genetics in their primary care practice. In addition, a case scenario is interwoven throughout the article to illustrate how evolving information in the primary setting can affect medical care and all of its associated factors (such as history gathering, testing, and consultation, to name a few.)

The themes from these articles serve as the basis for a consensus statement (Table 1) that consists of recommendations for the integration of genetics and genomics into pediatric primary care. The goal of this statement was to generate a blueprint for providing and fostering genetic and genomic literacy in primary care. The participants were mindful of the need to provide a blueprint to guide future initiatives and research opportunities; to guide policy decisions for medical education (at all levels from undergraduate, medical school, residency training, and continuing education); and to guide professional organizations in the integration of genetics and genomics into their societal initiatives. Although the participants were cognizant that they would not able to identify all the components and steps needed to advance genetics and genomics into primary care in a 2-day meeting, several major recommendations were developed (Table 1):

- Define how pediatric PCPs should use genetics and genomics in practice.
- Identify, develop, and provide the tools and resources that are needed to integrate genetics and genomics into primary care.
- Integrate genetics and genomics into primary care training at all levels.
- Provide an evidence base for optimal integration of genetics and genomics into primary care.

These recommendations do not imply that previous efforts in this field have been for naught; rather, the recommendations are intended to build on past efforts as we use the ever-increasing knowledge in this genomic era. The colloquium participants recognize, however, that important gaps exist in current knowledge. To strengthen clinical practice, inform policy-making, and improve child health in the era of personalized medicine, we must begin by providing evidence-based answers to the preliminary list of questions shown in Table 2. The colloquium participants fervently hope that the articles in this supplement and the consensus statement will guide future initiatives related to the integration of genetics and genomics into primary care.

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