Reflections on the Genetics in Primary Care Institute and Implications for the Future of Genetics in Pediatric Primary Care

A Project in Review

GPCI GENETICS IN PRIMARY CARE INSTITUTE

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GENETICS IN PRIMARY CARE INSTITUTE

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A cooperative agreement between the American Academy of Pediatrics and the Health Resources and Services Administration, Maternal and Child Health Bureau

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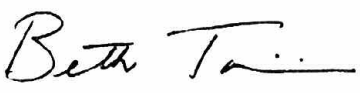
Thoughts From the Project Co-Medical Directors

The Genetics in Primary Care Institute (GPCI), a collaborative project of the American Academy of Pediatrics (AAP) and the Maternal and Child Health Bureau of the Health Resources and Services Administration (MCHB/HRSA), is pleased to reflect on its progress over the last 3 years. The GPCI sought to make the advances in genetics over the last 50 years, especially those since the completion of the Human Genome Project, relevant to the practice of primary care pediatrics. As noted on the GPCI Web site (www.geneticsinprimarycare.org), advances in infectious disease medicine revolutionized how medical care was delivered to children in the 20th century, with the advent of immunizations, antibiotics, and advanced diagnostic tools, and infectious disease medicine was thoroughly integrated into primary care. Advances in genetics will have a similar effect on the practice of pediatrics in the 21st century.

We now realize that the medical specialty of genetics represents the coalescence of genetics, genomics, and epigenetics. The challenging task of the GPCI has been to facilitate the translation of genetics information into resources that are relevant for the practice of primary care medicine. The potential for the prevention, diagnosis, and treatment of many genetic disorders is now at hand. As a result, developing the ability to use genetics to address the triad of prevention, diagnosis, and treatment has been our goal and is a key factor in making genetics relevant to primary care practice. Specifically, the GPCI has focused on helping to guide the pediatric primary care provider in maximizing evaluation and diagnostic processes related to genetic risk, becoming familiar with therapeutic modalities—such as the AAP health supervision guidelines—and assisting with the prevention of common and less common disorders.

This report highlights the advances that have occurred over the last 3 years in the integration of genetics into the areas of education, training, and practice improvement. As co-medical directors of the GPCI, we are often credited with the accomplishments of others. However, the work that has been done and the work that is still underway, as well as all the supporting documentation, could not have been accomplished without the incredible AAP team and all of our partners. We are honored to have been a part of this team. We invite you to review this report with an eye to the advances that have been made, to what is currently being accomplished, and to what could be achieved going forward. We hope that you share our enthusiasm for genetics in primary care, now and in the future. We believe that the efforts of the GPCI have "moved the needle" in integrating genetics into pediatric primary care practice. However, in doing so, we have uncovered more that will need to be done if the benefits of genetics in pediatric primary care are to be fully realized and sustained.

Robert A Saul, MD, FACMG, FAAP



Beth A Tarini, MD, MS, FAAP

About the Genetics in Primary Care Institute

The vision of the Genetics in Primary Care Institute (GPCI) was to increase the knowledge and skills of medical home primary care providers (PCPs) in providing genetics-based services. The GPCI was a 3-year cooperative agreement between the US Department of Health and Human Services - Health Resources and Services Administration - Maternal and Child Health Bureau (HRSA/MCHB), and the American Academy of Pediatrics (AAP). The agreement was structured around three overarching goals:

- To establish a community of learners who utilized quality improvement science to develop, implement, and evaluate strategies to improve PCPs' knowledge, practice, and attitudes regarding the provision of genetics-based services
- To implement a strategy to address systems and policy changes to accelerate the provision of genetic medicine
- To ingrain the practice of genetic medicine into the future PCP workforce by assessing and addressing postgraduate medical education and training

To achieve these goals, the GPCI developed a three-pronged approach involving a diverse set of educational offerings about genetics in primary care, facilitation on how to incorporate genetics into clinical primary care practice, and resources designed to meet pediatric residents' educational and training needs. A multidisciplinary project advisory committee oversaw all program activities.

This report describes the major activities of the GPCI, shares key results and the lessons learned, and offers insights regarding the future of genetics in primary care and strategies for overcoming persisting challenges. **The project has developed a foundation for how pediatric PCPs should think about genetic medicine; has formulated recommendations for operationalizing this foundation within the family-centered medical home; has increased knowledge through various educational offerings; and has gathered critical information about what resources should be developed to educate the future health-care workforce on how to integrate genetics into pediatric primary care.**

> All of the clinicians in our practice are extremely dedicated to making sure that our patients receive the best care possible, and we desperately want to make sure that our patients will have access to all of the benefits of personalized, genomic medicine.
>
> *—Participant in the GPCI Quality Improvement Project*

> All disease ultimately has a genetic basis.
>
> *—Joe McInerney, MA, MS, National Coalition for Health Professional Education in Genetics*

Establishing Relationships With Key Partners

Building relationships between other primary care, genetics, and public health organizations was a priority for the GPCI. **Through collaborative endeavors, the GPCI built upon existing projects and enabled the spread of information and the development of resources related to genetics in primary care.** The relationships established during the course of the 3-year cooperative agreement resulted in the tangible resources and activities described in this report. The emphasis on engaging multidisciplinary participation (including bringing to the table the perspectives of various stakeholders—such as family physicians, nurse practitioners, consumer advocates, and subspecialists) was a core tenet of each project undertaken by the Institute.

The GPCI Project Advisory Committee and various work groups included representatives from the following organizations:

- Academic Pediatric Association
- American Academy of Family Physicians
- AAP Committee on Genetics and Section on Genetics and Birth Defects
- American College of Medical Genetics and Genomics
- Genetic Alliance
- Association of Pediatric Program Directors
- Maternal and Child Health Bureau of the Health Resources and Services Administration, Genetic Services Branch
- National Association of Pediatric Nurse Practitioners
- National Coalition for Health Professional Education in Genetics
- National Society of Genetic Counselors
- National Institutes of Health

Additional partnerships have resulted in sharing resources between stakeholders to increase reach and cross-fertilization of ideas.

> **GPCI has been a close partner to Genetic Alliance throughout the cooperative agreement, with each organization cross-promoting one another's activities. Of late, we've collaborated more closely on the assessment of consumers' needs and how to incorporate effective patient education into the PCP training process.**
>
> *—James O'Leary, MBA, Chief Innovation Officer, Genetic Alliance*

> **It [the GPCI] has provided valuable resources that are easily (and freely) accessible. It [the GPCI] has allowed us to promote the value of genetic counselors as part of our strategic plans while also allowing us to help additional healthcare providers and patients.**
>
> *—Karin Dent, National Society of Genetic Counselors*

Collaborations with the National Coordinating Center for the Regional Genetic Service Collaboratives

The National Coordinating Center for the Regional Genetic Service Collaboratives (NCC) coordinates seven HRSA-funded Regional Genetic Services Collaboratives (RCs). Through a HRSA cooperative agreement with the American College of Medical Genetics and Genomics, the NCC and the RCs focus on bringing quality genetic and newborn screening services to local communities and on building bridges between public health organizations, primary care/medical home organizations, geneticists and other specialists, and families and consumers. The GPCI collaborated with the NCC and the RCs on a various projects, including the following:

In the national quality improvement project (QIP) conducted via the GPCI, participating primary care practices were paired with a genetics professional from the practice's region, who served as a mentor throughout the project.

GPCI staff participated in the NCC Medical Home Workgroup and the NCC Family Health History Workgroup, contributing feedback, information, and resources.

GPCI engaged in a collaboration between the Genetic Alliance and the NCC Medical Home Workgroup that involved assessing the needs of patients with genetic conditions, assessing the literature, and conducting qualitative research around the needs and experiences of parents with genetic conditions.

GPCI staff and faculty attended numerous in-person meetings of the NCC and the RCs to share resources and best practices identified and to facilitate connections between shared priorities regarding improved education for PCPs and provision of genetic services.

Collaborations With the Leadership Education in Neurodevelopmental and Related Disabilities Programs

The Leadership Education in Neurodevelopmental and Related Disabilities (LEND) programs are interdisciplinary leadership training programs administered by the Maternal and Child Health Bureau of the Health Resources and Services Administration and are federally funded through the Association of University Centers on Disability. The LEND programs "provide long-term, graduate level interdisciplinary training as well as interdisciplinary services and care. The purpose of the LEND training program is to improve the health of infants, children, and adolescents with disabilities. They accomplish this by preparing trainees from diverse professional disciplines to assume leadership roles in their respective fields and by insuring high levels of interdisciplinary clinical competence."[1]

GPCI staff participated in the LEND Genetics Workgroup, whose mission is to engage the LEND training network, and its partners, to play a key role in educating LEND trainees and healthcare professionals and increasing access to training provided by genetics professionals and other LEND interdisciplinary professionals on issues related to genetics and genetic counseling. Resources developed by the GPCI are included on the LEND Genetics Workgroup Web site and are used to educate LEND trainees. Additionally, AAP chapters that received Visiting Professorship Program awards were encouraged to partner with LEND programs in their state for the implementation of the Visiting Professorship Program in spring 2014.

Partnerships With Public Health Organizations and Federal Agencies

The GPCI partnered with the National Center on Birth Defects and Developmental Disabilities of the Centers for Disease Control and Prevention to hold the first medical education course for PCPs on genetics, titled *Dive Into the Gene Pool.* This milestone event, co-sponsored by the National Coalition for Health Professional Education in Genetics, brought together pediatric healthcare professionals from across the country for a 2-day continuing medical education conference in Chicago, Illinois, in August 2013.

GPCI staff members participate in the Federal/non-Federal Family History Work Group, facilitated by the National Human Genome Research Institute, to share resources and learn about best practices and new research in the field of family health history.

The blueprint for the GPCI emerged from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. One of the co-medical directors of the GPCI is the AAP liaison to the committee and serves as an active link between GPCI and national initiatives regarding genetics in public health and newborn screening. The GPCI and the AAP actively support the 50th Anniversary Campaign for Newborn Screening.

Since its inception, the GPCI has participated, via co-medical director Robert Saul, on the Inter-Society Coordinating Committee for Practitioner Education in Genomics. The committee was founded "to improve genomic literacy of physicians and other practitioners and enhance the practice of genomic medicine (that is, using an individual patient's genomic results in their clinical care) through sharing of educational approaches and joint identification of educational needs." These goals are accomplished by facilitating "interactions among professional societies that will enhance their efforts to increase the understanding and expertise of practitioners in applying genomic results to clinical care."[2] The AAP—through the initiatives of the GPCI—has been a recognized leader in this committee; has shared templates of survey instruments and information about overcoming barriers to the adoption of genomic medicine; and has actively participated in work group activities and discussions.

The GPCI has provided an opportunity for dissemination of some of NIH's activities and research areas of focus related to genetics and genomic medicine.

—Melissa Parisi, MD, PhD, FAAP, FACMG, National Institute of Child Health and Development, National Institutes of Health

Introduction

The number and frequency of advances in genetics have been unprecedented during the past 20 years. The mapping of the human genome and the development of hundreds of disease-specific genetic tests are just two examples of recent significant achievements. However, the **translation of advances in genetics into everyday clinical practice continues to be a challenge,** as is the case in many other areas of medicine.

Genetics and genomics play a major part in the health of the children of the US. Nearly 1 in 33 babies are born with a birth defect, and many of these defects are genetic.[3] Among pediatric inpatients, 71% have a genetic condition.[4]

Among the 4 million babies born annually in the US, about 1 in every 500-700 are found to have a treatable genetic condition.[5] The number of children with a diagnosed genetic condition has increased over the last 50 years owing to advances in screening, testing, and diagnostic techniques. Furthermore, new research suggests that an estimated 1 in 68[6] children have autism spectrum disorders, a diagnosis for which a genetic evaluation is recommended. Unfortunately, the number of genetics specialists has remained static in recent decades or, in some locations, even decreased during the same time period. Therefore, other healthcare providers, such as PCPs, must be relied upon for much-needed genetics services. **PCPs are well-suited to address genetics because they develop long-term meaningful relationships with their patients, they care for their pediatric patients as well as interact with multiple generations of the patients' families, and they are well-positioned to coordinate care between specialists and hospitals.** It was against this backdrop that the AAP applied for, and was subsequently awarded, a competitive grant to establish the GPCI in 2011.

Incorporating genetics into primary care involves a shift of paradigm in medical thinking. It is not simply the "dermatologic approach" of matching physical characteristics to syndromes in a book or diagnosing disease through identification of a gene mutation. It involves thinking about the genetic makeup of each individual in every encounter to develop differential diagnoses for disease or for preventive counseling. In this scenario, child health providers will need to incorporate a new approach to primary care, with emphasis on certain skills, and also will need to be reeducated and updated on new knowledge in genetics.

—Cheng TL. Primary care pediatrics: 2004 and beyond. Pediatrics. 2004;113(6):1802-1809

We can tailor care if we know someone has [a condition] based on changes in certain genes or certain environmental influences.

—Louanne Hudgins, MD, FACMG, Lucile Packard Children's Hospital

Laying the Foundation

An important first task of the GPCI was to survey the available knowledge regarding genetics in pediatric primary care. Understanding the starting point was critical for deciding how the project should proceed and for identifying areas that might be outside the scope of the project but that would nonetheless need to be addressed in the future. To accomplish this task, the GPCI conducted various activities, including preparing a summary of similar past projects; interviewing key stakeholders on what they felt were the most influential events to date and the most pressing challenges; systematically reviewing the barriers to integrating genetics into primary care; conducting a needs assessment survey of a subset of AAP members; and, ultimately, conducting a national survey to investigate practice patterns around collecting family health histories. The key findings from each of these activities are described below.

Review of Previous Genetics Education and Implementation Projects

Four projects that investigated topics similar to those addressed by the GPCI were carefully reviewed via published literature and interviews with the project leaders. This review highlighted valuable lessons, regarding both successes that the GPCI could emulate and challenges to be avoided. A few of these lessons, which were subsequently incorporated into the GPCI-QIP, are as follows:

- A trusted, readily accessible clearinghouse for information for practicing clinicians should be created.
- Follow-up data should be collected to understand whether the changes implemented in practice behavior as a result of the project are sustainable.
- Education to PCPs needs to be provided by primary care experts as well as by geneticists.
- Case-based learning is valuable.
- Patients' perspectives on genetics in pediatric primary care should be assessed.

The review of the past projects also highlighted areas of need that did not necessarily fall under the purview of GPCI but are important for the continued integration of genetics into primary care medicine, such as the need to:

- focus on resources and education around specific common disorders, such as attention deficit hyperactivity disorder and autism, that have a genetic basis
- understand how genetic literacy of healthcare providers and the public and current culture impact the integration of genetics into practice
- investigate how to best use electronic health records (EHRs) to incorporate genetic and genomic information at the point of care and facilitate the integration of genetics into practice

Stakeholder Interviews

In assessing the state of genetics in pediatric primary care at the outset of the GPCI, seven national thought leaders and experts in genetics education and practice were interviewed regarding their thoughts on (1) the evolution of genetics in primary care, (2) barriers to incorporating genetics into primary care, (3) key competencies needed by PCPs to incorporate genetics into their practices, (4) moving genetics in primary care forward, (5) the role of the consumer, and (6) actionable items for the GPCI. Key points raised by

the interviewees included the following:

- Genetics in primary care is a moving target because of the rapid progress occurring in genetics.
- PCPs can supplement existing genetics services because there will never be enough geneticists:

  There has been an explosion in diagnostic technology and treatments, with newborn screening there are more conditions that can be found, but the reality is that there are not enough geneticists out there to perform services, so PCPs need to fill that gap.

- However, an increased reliance on PCPs means that they will need help to navigate the "genetics minefield":

  [The PCP lacks] fundamental knowledge about what things mean and what to do, and people are uncertain of where to turn to for specialty help.

- Defining key competencies and the PCP's role in genetics is crucial, and integrating knowledge into training is crucial:

  If you put genetics information on board exams that is a way to push, if you need to know some of this stuff to maintain your certification that is... pretty good.

- Championing the cause is critical to integrating genetics into primary care.

  The AAP has a unique opportunity to take leadership in this.

Exploring PCPs' Perceptions About Barriers to Providing Genetics Services: A Systematic Review of the Literature

The GPCI systematically reviewed the literature to identify what PCPs perceived as being barriers to providing genetics services. Published articles from 2001 to 2012 were considered for inclusion, and 38 publications were selected from a pool of over 4,000 that met the inclusion criteria. The 38 publications revealed 311 unique perceived barriers, which were classified into four distinct groups: (1) barriers due to lack of knowledge and skills, (2) ethical, legal, and social barriers, (3) barriers related to healthcare delivery systems, and (4) barriers resulting from the lack of scientific evidence and professional guidelines regarding genetic testing and risk assessment. A quality assessment was also conducted to ascertain the level of bias of the quantitative studies that were reviewed (n = 25) and the overall quality of the qualitative studies that were included in the review (n = 13).

Within each group, the barriers were ranked according to the number of times they were cited across the 38 publications. The systematic review generated the following results:

- Barriers most frequently mentioned by PCPs included a lack of knowledge about genetics and genetic risk assessment, concerns about patient anxiety, a lack of access to genetics services, and a lack of time.
- A similar review conducted in 2001 cited similar barriers; however, since that time, there had been a major shift, an increase in perceived importance and relevance of genetics in primary care.
- PCPs indicated that genetics impacted their day-to-day practice and that **they would benefit from targeted educational resources and point-of-care tools for assessment of genetic risks, tools for patient communication, and for appropriate utilization of genetics services.**

Information from the systematic review should be used to formulate multipronged interventions aimed at improving genetics services in primary care; interventions that consider these barriers might be more effective.

Genetic Services in Primary Care Pediatrics: A Survey of Physicians in the AAP Quality Improvement Innovation Networks

To identify the current practices of PCPs regarding patients with genetic conditions, the GPCI conducted an online needs assessment survey of PCPs' attitudes toward genetic medical care and their choices regarding the taking of family health histories.[7] This survey was completed by 88 PCPs associated with the AAP Quality Improvement Innovation Networks (29% response rate). The survey results indicated areas where PCPs were incorporating genetics into their practices and highlighted opportunities for improvement:

- 100% agreed or strongly agreed that taking a family health history is important, and 32% agreed or strongly agreed that they currently gather at least a three-generation history.
- 49% agreed or strongly agreed that they feel competent in providing healthcare related to genetics and genomics.
- 13% reported discussing the risks, potential benefits, and limitations of genetic tests with patients.
- 86% of respondents reported ordering genetic tests three times or fewer annually

Table 1: Attitudes and Practices in Family History Taking by Primary Care Pediatricians

Item	N	Strongly Agree (%)	Agree (%)	Disagree (%)	Strongly Disagree (%)
Taking a family health history is an important part of the assessment of an individual's [or patient's] predisposition to disease	87	**57 (66)**	30 (34)	—	—
I gather a minimum of three generations of family health history information for all of my patients	86	10 (12)	17 (20)	**50 (58)**	9 (10)
When collecting a three generation family history, a barrier that I face is that patients are unable to provide accurate/complete information.	87	34 (39)	**51 (59)**	2 (2)	—

Item	N	Always (%)	Most of the time (%)	Some of the time (%)	Rarely (%)	Never (%)
For each of the following visit types, how often do you obtain or update a family health history for a patient?						
At first visit [either acute or health maintenance]	87	**43 (49)**	33 (38)	10 (11)	1 (1)	—
At health maintenance visits	87	14 (16)	**35 (40)**	30 (34	7 (8)	1 (1)
When a specific health problem arises	85	23 (27)	**33 (39)**	25 (29)	4 (5)	—
How often do you construct a pedigree (eg, diagram of family members' relationships) with this information?	86	2 (2)	2 (2)	8 (9)	**44 (51)**	30 (35)

Most frequent answer is bolded

Periodic Survey of AAP Fellows

Aimed at understanding the current state of genetics in pediatric primary care, a national survey, in the form of a self-administered questionnaire, was sent to a representative sample of AAP members in the winter and spring of 2014. The purposes of the survey were (1) to explore practice patterns with respect to the collection of family health histories and to understand factors affecting the gathering of this information and (2) to assess the respondents' perceptions of and behaviors regarding genetic testing and interpretation/evaluation of the results. Preliminary data from this first nationally representative survey of pediatricians regarding genetics in primary care revealed the following:

- Nearly 95% of respondents collect family health history information from their patients, and 80% of history information is collected in the exam room and entered into the EHR.
- More than two-thirds of respondents have a policy in place for collecting family health history. Respondents typically ask open-ended questions (41%) or use a standardized disease checklist (49%) to collect family health history.
- Nearly 40% of respondents sometimes or rarely take family health histories from their established patients, and 27% of respondents sometimes or never update this information at annual health maintenance visits.
- Nearly two-thirds of respondents reported that inadequate time during a typical office visit is a moderate or significant barrier to collecting or updating family health histories, whereas only 10% reported that lack of time was not at all a barrier.
- Depending on the patient encounter type, most respondents collect family health history for a range of disorders: for example, disorders relevant to clinical findings (95%), disorders relevant to the child's age-based health (78%), and adult-onset disorders not immediately relevant to the child's age-based health (57%).

Ninety-one percent of the respondents agreed or strongly agreed that genetic testing can provide important information regarding recurrence risks for other family members, and nearly 74% of respondents agreed or strongly agreed that there are situations in which it is the primary care pediatrician's role to order genetic tests. However, less than one-third of respondents are confident in their ability to interpret genetic testing results. Survey respondents reported a lack of training in identifying genetic risks, choosing appropriate tests, and interpreting the results. Over two-thirds of survey respondents are moderately or very interested in seeking continuing medical education regarding genetics in primary care.

Changing Practice

GPCI-QIP

Overview

The delivery of high-quality pediatric care requires that PCPs be knowledge about medical genetics and play a role in providing genomics services. The family health history provides a basis for identifying risks and concerns based on the interplay between a child's genetics and his or her environment, and is a core component of preventive care. Ongoing care and management of patients with identified genetic conditions is equally important.

Over a year-long quality improvement initiative, the GPCI-QIP mobilized 13 diverse primary care practices (in 11 states) that used quality improvement science to test, implement, and evaluate strategies to improve knowledge, practice, and attitudes regarding the provision of genetics services. The GPCI-QIP focused on improving the identification and assessment of children with genetic conditions, as well as improving the management of their care.

The assessment of children with genetic conditions begins with collection of an adequate family health history and physical examination during health supervision visits for all patients, starting at birth and continuing to age 21. Additionally, practices worked to ensure that patients diagnosed with or at risk for a genetic condition received appropriate follow-up, including referral or re-referral to a geneticist, and that appropriate services were provided to them. This project also focused on improvements in systems for tracking children with genetic conditions to ensure that appropriate follow-up, communication, and care coordination took place.

Project Aims The aims of the project include the following:

1. To improve the assessment and identification of genetic conditions for all patients from birth to age 21, as part of health supervision visits. Specific goals included the following:
    - Family health histories are created or updated at health supervision visits, using the family health history components defined by the project, for 90% of patients.
    - Current family health histories are discussed with 90% of patients and their families.
    - 90% of patients with a positive family health history or identified clinical symptoms about a genetic condition have documentation in their charts that follow-up or a plan of care was discussed with the patients and their families.
    - 90% of practices plan for and accommodate patients' special needs at office visits.
    - 90% of practices have a system for recalling patients who are due or overdue for preventive services.
    - 90% of practices have a referral-tracking mechanism.
    - 90% of practices have a process or written protocol for use of a standardized family health history tool.

2. To improve the care and management of patients aged 0–21 years with defined genetic conditions. Teams will specifically work to improve the percentage of such patients who have the following:
    - Documentation that health supervision guidelines have been followed (90%)
    - Up-to-date health supervision visits (90%)
    - Documented next steps and follow-up, as appropriate (90%)
    - Documented emergency plans, if applicable (90%)
    - Documentation that genetic services have been offered, at least initially (90%)
    - Documentation that transition of care to adult providers has been discussed at least annually for patients age 12–21 years (90%)
    - Documented that palliative care has been discussed at least annually, if applicable (90%)

Patient Demographics

Practice type

- Independent practice (n = 9)
- Affiliated with a university or medical school (n = 2)
- Hospital-affiliated practice (n = 1)
- County public health department or clinic (n = 1)
- One family medicine practice participated.

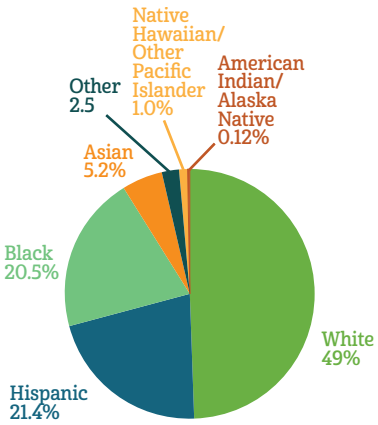
Setting

- Inner-city urban (n = 3)
- Suburban (n = 6)
- Non-inner-city urban (n = 4)

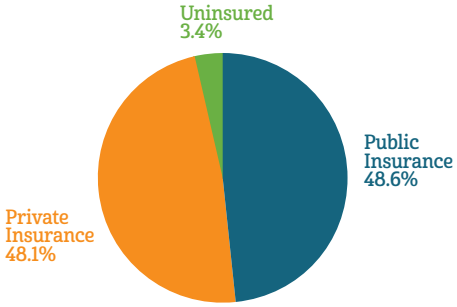
Other

- The number of pediatricians in the practices ranged from 1 to 18 (excluding residents)
- Practices saw 1,500–24,000 pediatric patients annually, with an average of 10,000
- All practices used an EHR

Race/Ethnicity



Insurance Status



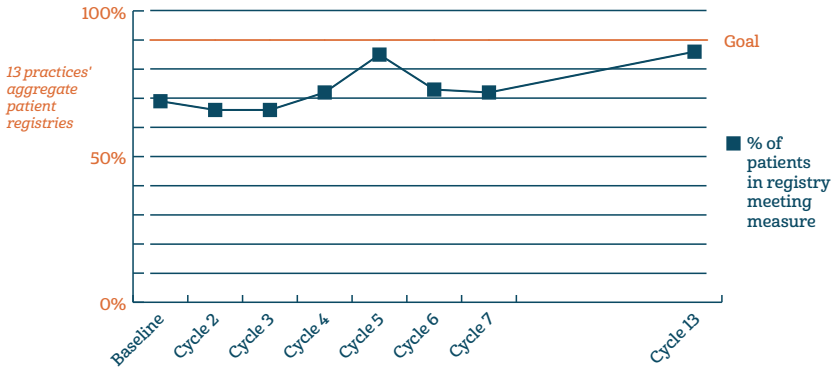
Results

The most significant result of the GPCI-QIP was improvement in the collection of detailed family health history information as part of health supervision visits; 100% of the practices were actively collecting comprehensive information 6 months after the project intervention. By including family health history in health supervision visits, providers could use the obtained information to improve screening of patients with genetic conditions or at risk for adverse health events, and could improve delivery of care by using algorithms to guide risk assessment and clinical action, ordering genetic tests, and referring patients to a geneticist or genetic counselor.

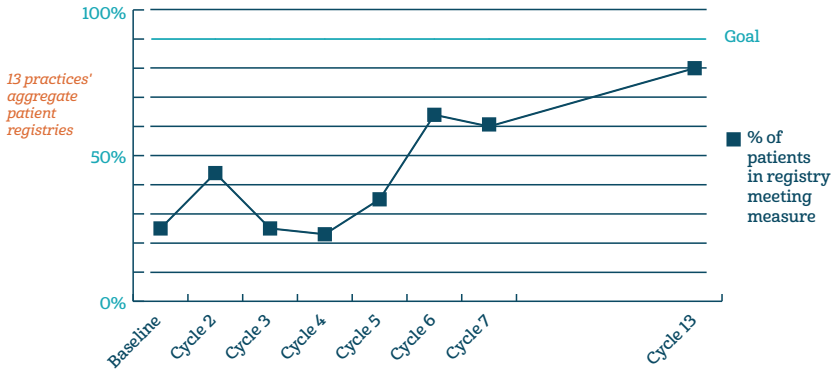
Care delivery was also significantly improved by the development of a patient registry of practice-level populations of children with genetic conditions. This registry also effectively improved the quality of care management through the use of health supervision guidelines, the creation of care plans, the documentation of emergency care plans, the development of transition policies, and more.

> **Success is consistency—we've all bought into the importance, that we are consistent in handing out the [family health history] forms and following up on the information. We need to keep working to capture the data, and need to build the knowledge to interpret the data.**
>
> *—GPCI-QIP participant*

Age-appropriate Health Supervision Visits

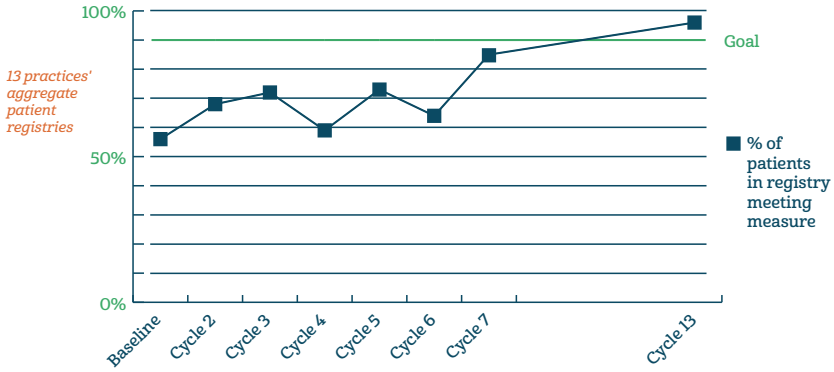


Documented Next Steps/Plan of Care in Registry



These graphs show aggregate data from the practice-level registries of a patient population with diagnosed genetic conditions prior to the intervention (baseline), during the intervention (cycles 2–7), and 6 months after the intervention (cycle 13).

Use of Health Supervision Guidelines for Children with Genetic Conditions



The success of targeted strategies to improve the primary care of children with genetic conditions requires a multifaceted approach to improving healthcare processes and systems. Best practices identified through the GPCI-QIP include the following:

- Engage information technology experts in patient registry functions and modifying family health history screens in the EHR.
- Obtain buy-in from clinic staff and providers regarding the utility of family health history.
- Implement small tests of change and obtain team members' feedback on the process.
- Establish office-wide procedures and policies for patient management.
- Cultivate relationships with local genetics professionals.

Practice has a standard process (written or not written) for	Pre-QIP	Post-QIP	Δ
Tracking genetic referrals made	38%	85%	47%
Identifying patients with a defined genetic condition	23%	77%	54%
Tracking children with a defined genetic condition	23%	77%	54%
Identifying genetic patients who are behind schedule for preventative services	23%	69%	46%
Contacting genetic patients who are behind schedule for preventative services	38%	69%	31%
Discussing transition to adult care with genetics patients by age 12	15%	62%	47%
Co-managing care of genetics patients	8%	31%	23%
Obtaining emergency plans from specialists	15%	31%	16%

Practices offered varying responses of how they described their success in project participation. Successes could be grouped into five categories:

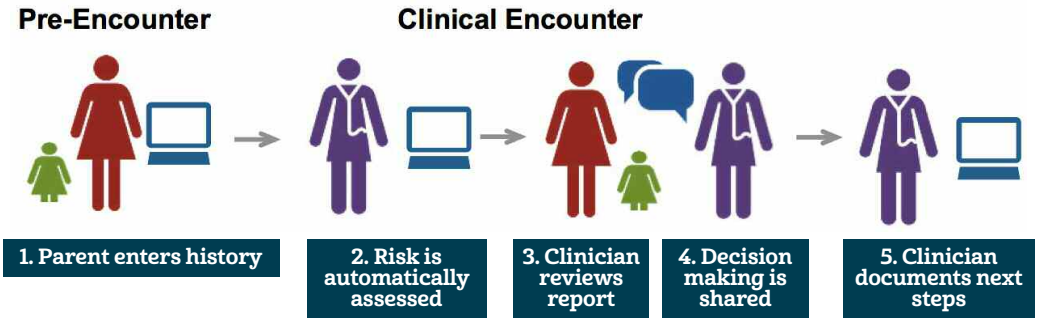
1. increased knowledge
2. measurable change
3. consistency in implementing change
4. improved outcomes for patients
5. commitment to continuous improvement

A Toolkit to Improve Care for Pediatric Patients with Genetic Conditions in Primary Care

Throughout the action period, practice teams used a robust Change Package, containing resources, information, and various tools to test in their clinics. As a result of the project, vetted tools and best practices were compiled into a comprehensive toolkit. This resource provides practice improvement strategies and resources tested by the practice teams to integrate genetics into the medical home and will be promoted widely to an audience of pediatric primary care clinicians and organizations.

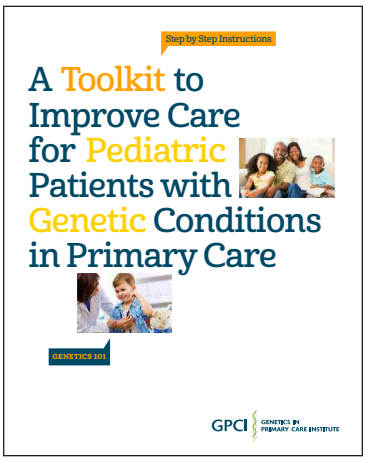
Developing Innovative Family Health History Tools

The GPCI, in collaboration with the National Coalition for Health Professional Education in Genetics, March of Dimes, Genetic Alliance, and Partners HealthCare developed an electronic, patient-entered family health history tool for pediatric PCPs. The tool supports the provider in conducting a family health history risk assessment and genetic evaluation. It identifies additional evaluation services, preventative services, and personalized management for children who are at increased risk for certain health conditions owing to their family health history. The tool was developed on the basis of an extensive review of pediatric family health history tools, a needs assessment, published literature, and professional guidelines. The tool collects information on 35 conditions; most of the conditions are genetically complex, but a few single-gene disorders are included.



GPCI has laid a foundation of practical information that PCPs need to know to integrate genetics into their practice. By focusing on how to institute incremental change of practice through clear, concrete steps, they were able to make the daunting field seem accessible. In addition, their work to show the impact of connecting PCPs to genetics expertise (in partnership with the Regional Genetics Collaboratives) was an important first step in determining the future relationship of those two disciplines.

—James O'Leary, MBA, Chief Innovation Officer, Genetic Alliance



Electronic Family History Tool: How It Works

Collects structured patient (or parent)–entered data about family health history

- In initial testing, an average of 9 minutes was required to complete the questionnaire
- Is built according to Health Level 7 standards and is interoperable with US Surgeon General's My Family Health Portrait and other family health history tools

Automatically assesses risks for 35 health conditions associated with family health history:

- Lists available interventions for children at increased risk, such as screening or diagnostic tests, additional evaluation or referral, modified management, preventive services, or counseling and education

Builds a medical pedigree that includes first-, second-, and third-degree relatives and that can be updated at any time

Applies algorithms developed from the guidelines of professional organizations, including the AAP and the American College of Medical Genetics and Genomics, and from published literature

- Presents the provider with clinical decision support in the form of actions for consideration
- Creates a personalized report that can be included in the medical record and documentation of the encounter

The development of the pediatric family history tool and creation of quality improvement projects for genetics is excellent.

—Marc Williams, MD, FAAP, FACMG, Geisinger Health System

Integrating Genetics Into the Health Supervision Visit Through *Bright Futures*

Bright Futures: Guidelines for Health Supervision of Infants, Children, and Adolescents supports the preventive aspects of the family-centered medical home. This publication is also considered the premier source of anticipatory guidance information for the pediatric healthcare community and is the standard for providing preventive pediatric healthcare as defined in the Affordable Care Act. The third edition of *Bright Futures* suggests that "interval history may be obtained according to the concerns of the family and the health-care professional's preference or style of practice, or by using an age-specific-visit parent questionnaire."[8]

The GPCI recognizes that the use of family health history to inform health promotion, disease prevention, and optimal patient outcomes must be more closely examined. Therefore, the GPCI seeks to enhance the inclusion of genetic medicine principles in the next version of *Bright Futures* health supervision guidelines. For future iterations of *Bright Futures,* the GPCI has suggested incorporating additional evidence-based information about the importance of collecting family health history information as it becomes available. The GPCI suggests that a brief, yet comprehensive, family health history screening tool with concise, targeted language be used at all new patient visits, followed by an annual review of family health history information to assess relevant changes in the health status of family members; this approach was shown to be practical in the national QIP (see page 14).

Patients expect you to explore what's going on with their family. It helps us understand what things we should be looking for when we know that history.

—Timothy Geleske, MD, FAAP, North Arlington Pediatrics

Providing Education

One of the essential missions of the GPCI is to educate a broad audience of pediatric PCPs, healthcare professionals, and other stakeholders about how to incorporate genetics into pediatric primary care and to provide technical assistance in accomplishing that goal. A wide variety of modalities is necessary to meet the needs of individual learners. Therefore, the GPCI has provided educational offerings in various formats, including online, in-person, written, and video. To fill a previously identified gap, the GPCI has developed an educational Web site to serve as a clearinghouse for PCPs to obtain information and resources related to genetics and genomics. Over the course of the 3-year project period, technical assistance was provided to almost 7,000 individuals representing healthcare organizations, state maternal and child health agencies, community-based organizations, and more.

It will be important to know about the underlying genetic basis for all the conditions, rather than the rare conditions that we're able to diagnosis now, because that's where the technology is moving.

—Louanne Hudgins, MD, FACMG, Lucile Packard Children's Hospital

The GPCI Web Site

Launched in March 2013, the GPCI Web site (www.geneticsinprimarycare.org) has been commended as being a user-friendly, trusted source of information about incorporating genetics into primary care. Since its launch, the Web site has had more than 15,000 visits and more than 38,000 page views.

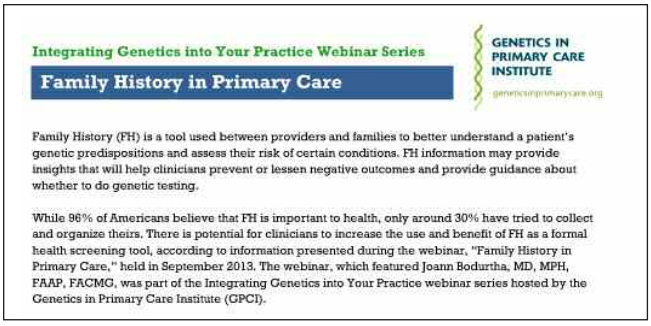
The Web site contains the following sections:

- Genetics in Primary Care, which includes a glossary of key terms, testimonials about the importance of genetics from members of the GPCI program advisory board, and various references and resources
- Genetics in Your Practice, which includes subsections on the importance of integrating genetics into primary care; family health history; genetic testing; quality improvement; ethical, legal, and social issues; financing and payment; when to refer; and patient management guidelines
- Provider Education, which includes archived webinar series, information about the Visiting Professorship Program, and other learning opportunities offered by GPCI partners

Educational Webinars

A webinar series called Time Out for Genetics was launched in March 2012. Ten topics were covered:

- Integrating Genetics in Primary Care—Why Does It Matter?
- Building an Accurate Family History, Constructing a Pedigree—An Overview for Primary Care
- Ordering the Right Tests—Genetics in Primary Care
- Genetics Evaluation, Referrals, and More—What to Do Next
- Myths of Primary Care Providers and Patients/Families Regarding Genetics—Setting the Record Straight
- Heard About Genetic Counseling? What Does It Mean for You, Patients, and Families?
- Genetics and Coding: What the Primary Care Provider Needs to Know
- Top 10 Genetics Resources for Pediatric Primary Care Providers
- Genetics Across the Lifespan—Putting It All Together
- Epigenetics—What Your Patients Are Asking, What You Need to Know



Integrating Genetics into Your Practice Webinar Series

Family History in Primary Care

GENETICS IN PRIMARY CARE INSTITUTE

Family History (FH) is a tool used between providers and families to better understand a patient's genetic predispositions and assess their risk of certain conditions. FH information may provide insights that will help clinicians prevent or lessen negative outcomes and provide guidance about whether to do genetic testing.

While 96% of Americans believe that FH is important to health, only around 30% have tried to collect and organize theirs. There is potential for clinicians to increase the use and benefit of FH as a formal health screening tool, according to information presented during the webinar, "Family History in Primary Care," held in September 2013. The webinar, which featured Joann Bodurtha, MD, MPH, FAAP, FACMG, was part of the Integrating Genetics into Your Practice webinar series hosted by the Genetics in Primary Care Institute (GPCI).

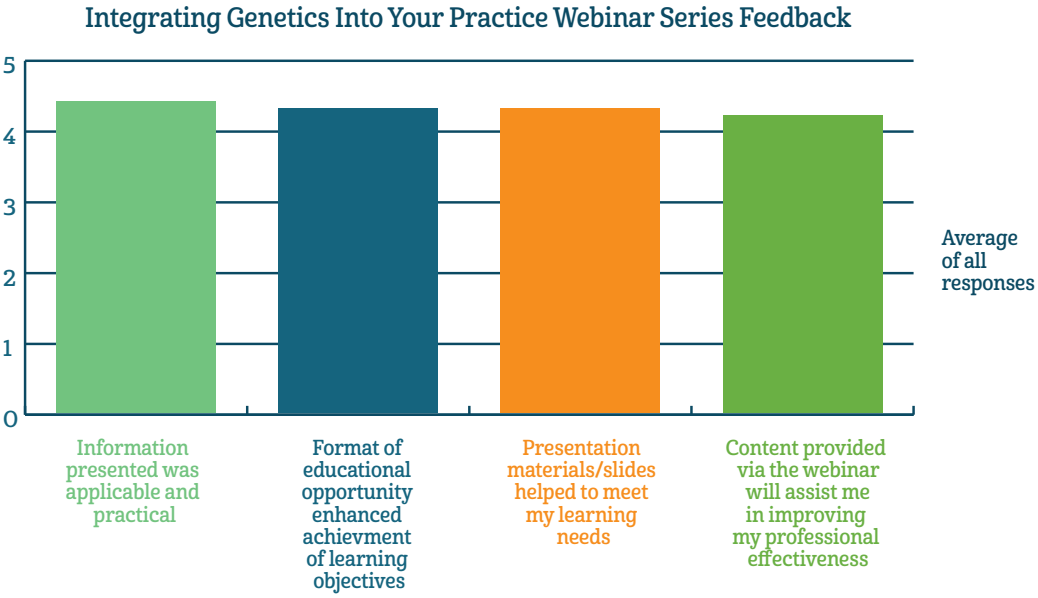
Webinar participants could question experts during the live webinar and were provided with slides from the presentation and a fact sheet for reference. A total of 3,414 people registered for the series, and 1,438 attended the live sessions. Feedback was collected through an online survey completed by 636 of the participants. At least 80% of the survey respondents gave positive feedback on the format, content, and audio and visual quality of the webinars. More than two-thirds of the respondents (69%–86%) indicated that the webinar series would improve their professional effectiveness. The series provided a forum for busy professionals to increase their knowledge about genetics in the primary care setting.

Very well done to help physicians and others start to think about future risks for patients, now that we know so much more about genes and their impact.
—Webinar participant

On the basis of the success of the Time Out for Genetics series, GPCI launched a second webinar series in September 2013 called Integrating Genetics Into Your Practice, which consisted of six 1-hour sessions:

- Family History in Primary Care
- Overview of Genetic Testing and Screening
- Genetic Red Flags in Well-Checks
- Genetic Testing in Primary Care
- Genetics Counseling in Primary Care
- Co-Management and Transitions for Patients With Genetic Conditions

On the basis of feedback from the first webinar series, the topics in the second series were more geared toward the primary care setting and less toward traditional genetics. A total of 2,074 people registered for the second series, and 955 attended the live sessions. Feedback was collected through an online survey; 75% of the respondents ranked the webinars as very good or excellent, and 46.5% said they would change their practices to provide better patient care as a result of participating in the webinar.



Dive Into the Gene Pool—A Conference for Pediatric PCPs

Webinars were just one of the educational offerings of the GPCI. A one-and-a-half-day workshop titled Dive Into the Gene Pool: Integrating Genetics and Genomics Into Your Pediatric Primary Care Practice was held in August 2013 in Chicago. The workshop was dedicated to providing PCPs with the knowledge, skills, and tools needed to integrate genetics into their everyday practice. The program included sessions on family health history, genetic red flags, genetic testing, communication with teens regarding genetics, managing referrals, genetics in the patient-centered medical home, and genetic components of common pediatric conditions. Participants were eligible to receive continuing medical education credits from their professional organizations. Seventy-five people attended the conference and committed to modifying their EHRs, developing a system to collect family health history following birth, updating the history annually for each patient, improving informed consent for genetic testing, and ordering genetic tests when appropriate.

Dive Into the Gene Pool PediaLink Course

An online continuing medical education course entitled Dive Into the Gene Pool was developed to provide practicing pediatricians with an opportunity to learn more about genetics in primary care and was offered via PediaLink. PediaLink is the AAP platform for lifelong e-learning and provides personalized education for healthcare professionals. The course includes relevant case-based content aimed at achieving the following learning objectives:

- Recognition of the role of the PCP in the integration of information about genetics, genomics, and epigenetics into practice.
- Application of effective communication strategies for interpreting and acting upon genetic test results.
- Describing the role of family health history as a tool for disease prevention, diagnosis, and management in the primary care setting.

Visiting Professorship Program

In 2014, the GPCI announced its Visiting Professorship Program. Implemented in collaboration with AAP chapters, the program awards funds to provide education about the importance of genetics in the pediatric primary care setting and to provide practical tools and strategies for improving everyday patient care. The program content focuses on the following areas:

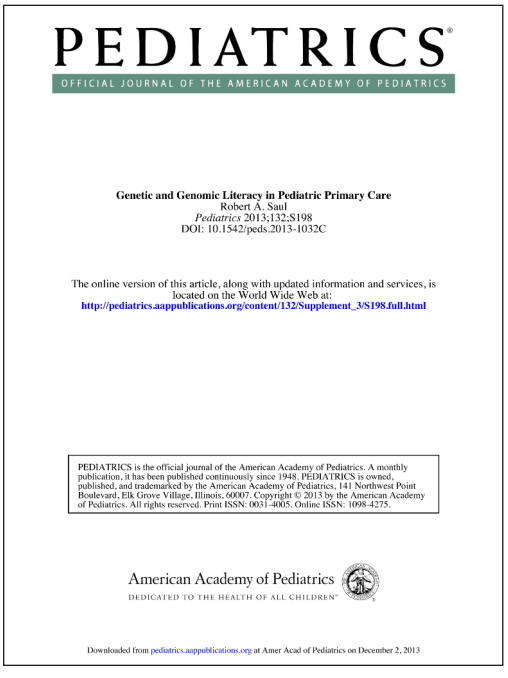
- Integrating genetics into primary care through the collection of family health history
- Communicating with patients about genetic risk
- Identifying indications for referral to a geneticist or genetic counselor, including red flags, family health history, and abnormal newborn screening results
- Assessing the role of the primary care clinician in the care of children with, or at risk for, a genetic condition

Visiting professorships were awarded to the AAP chapters in Alabama, California, Kansas, Missouri, Nevada, and New York. These six chapters collaborated with local healthcare organizations and LEND programs to hold a total of 20 educational sessions covering various genetics topics; the sessions, which offered numerous learning opportunities, included grand rounds, webinars, symposia, and lunch discussions at healthcare institutions.

Most of the participants (55%) at an educational session in New York* said that the knowledge they gained would very much affect their practice, and more than 70% said the session would increase their ability to review the medical and developmental features of common genetic disorders; to review the natural history, developmental trajectory, and prognostic indicators for these disorders; and to discuss projected interventions that might improve outcomes among individuals with these disorders.

Genetic Literacy in Pediatric Primary Care Colloquium

The Genetic Literacy in Primary Care Colloquium was held in fall 2012. The goal of the colloquium was to provide context for the delivery of genetics-related services in daily pediatric primary care practice, including prevention, diagnosis, and management. An executive summary and five articles were subsequently published in a supplement to Pediatrics in December 2013.[9–14] The colloquium topics were as follows:


PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Genetic and Genomic Literacy in Pediatric Primary Care
Robert A. Saul
Pediatrics 2013;132;S198
DOI: 10.1542/peds.2013-1032C

The online version of this article, along with updated information and services, is located on the World Wide Web at:
http://pediatrics.aappublications.org/content/132/Supplement_3/S198.full.html

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

- Genetic and Genomic Literacy in Pediatric Primary Care
- Family History in Primary Care Pediatrics
- Whole-Exome/Genome Sequencing and Genomics
- Epigenetics and Primary Care
- Genetic Literacy and Competency
- Primary Care and Genetics and Genomics

**At the time this report was developed, the VPPs were still underway and limited evaluation data was available*

Consensus Statement

The consensus statement represents a collaborative effort of all the colloquium participants. The goal 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 blue print 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.[9]

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 health histories and provide tools to facilitate the collection of 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.

Ingraining Genetic Thinking Into the Future Workforce

One goal of the GPCI project was to assess the education and training needs of the future PCP workforce in regard to integrating genetics into primary care. For physicians, medical education consists of medical school, residency, and oftentimes a fellowship. There is wide variation in how genetics is covered at each of these stages, and, unfortunately, the genetics content is taught from a traditional genetics framework, further perpetuating the notion of genetic exceptionalism. To understand the current state of genetics education in pediatric residency programs, the GPCI developed a 29-item needs assessment survey that was administered to all pediatric residency program directors in the United States. Forty-seven directors completed the survey (response rate 24%). After the survey was completed, five pediatric residency directors were interviewed to obtain further insights on the aggregate survey results.

Making resources available to residents is a good thing. Finding faculty who can oversee/implement/evaluate the use of the materials is another [challenge].

—Pediatric residency program director #1

Needs Assessment

The pediatric residency program directors represented a range of institution types. The size of the programs varied from small (5 residents) to large (127 residents), with an average size of 48. Most directors represented a university hospital system (54%), and most were in an urban setting (67%).

- Roughly two-thirds of survey respondents indicated that their program has an elective rotation in genetics; only 1 in 10 indicated that their program has a mandatory rotation in genetics.
- Because most residents do not participate in a genetics rotation, they receive instruction via other routes, usually through grand rounds. Eighty-three percent of respondents indicated they have MD geneticist faculty for genetics instruction.
- Respondents expressed interest in having targeted tools or resources for their residents, with two topic areas garnering the most interest: (1) genetic evaluation of children with cognitive or developmental delays and (2) recognizing genetic red flags in clinical evaluations.
- Crowding of the curriculum is a major barrier to incorporating genetics, now and in the future, into residency training.
- Respondents preferred that resources be designed for residents, as opposed to for faculty as training resources, and requested content in formats such as short online tutorials, videos, or PowerPoint slides.

Exploding information in the field [is] hard to manage and master. Residents need help with basics, and other specialized areas, such as when to refer and tap into specialty expertise

—Pediatric residency program director #2

The GPCI gathered input from pediatric residency directors from around the country on the presence, or lack, of genetics within their curriculum. Genetics is largely being viewed and taught from a traditional specialist approach. The survey results and the interviews showed that this barrier is significant and that more work will be necessary to convince PCPs that this is an issue that they must face because it can affect their patients.

Case Series Development

On the basis of a the results of the needs assessment, a work group determined that the development of an interactive case series for the continuity clinic, which is a part of resident training programs, would effectively address the need to educate residents about genetics in primary care. The series, entitled Think Genetics!, includes nine genetic topics relevant to the primary care pediatrician:

- The Importance of Family History in Pediatric Primary Care
- Tracking and Monitoring Patients With Genetic Conditions
- A Child With Short Stature
- Genetic Evaluation of Children With Cognitive/Developmental Delays, Possible Autism

- Recognizing Genetic Red Flags in Clinical Evaluations
- Genetics and Common Disorders in Pediatrics
- Newborn Screening
- Genetic Testing: The Nuts and Bolts
- Genetic Testing: The Bells and Whistles

All case reports were prepared by physicians and genetics experts, were aligned with pediatrics milestones, and were accompanied by resources both for professionals and for patients and caregivers, a glossary, and an evaluation tool. An extensive peer review process was used to ensure the validity, utility, and consistency of the cases.

The case series focuses on a wide range of clinical topics that are encountered in pediatric primary care and that require the resident to "think genetics" to adequately diagnose and treat patients in the continuity clinic. Without evaluating children through the lens of genetics, a pediatrician could miss critical pieces of the puzzle when caring for patients. Through these case exercises, residents will learn how genetics can be seamlessly incorporated into a primary care practice.

Case Objectives

Upon completion of the Think Genetics! case series, residents will be able to:

- recognize the role of genetics, genomics, and epigenetics in health and disease
- identify the role of the primary care clinician in the identification and management of patients with genetic conditions using family health history, expert referrals, and genetic testing
- describe the importance of constructing a problem-based family health history and three-generation pedigree when considering a diagnosis, prevention, or treatment plan in pediatric primary care
- apply a wide range of available resources, technologies, and tools that promote early identification, referral, and management of patients with genetic conditions

Summary and Looking Ahead

Moving the Needle

Battling genetic exceptionalism was a significant challenge prior to and during this project. **Raising awareness of the relevancy of genetics in primary care and connecting clinicians to existing resources will clearly need to be the continued focus until there is more mainstream acceptance of the PCP's role in thinking, and practicing, genetically.**

The needs assessments conducted throughout the project revealed that the uptake of genetic risk assessment and genetic services in primary care has been limited over the past decade and that several barriers—both systems-related challenges and preconceived "myths"—prevail. The GPCI made a concerted effort to address existing gaps and barriers, and in this process also encountered additional challenges and opportunities. Gaps that have been identified must continue to be resolved through continued spread of information and targeted implementation strategies across systems and health care professionals.

> The Genetics in Primary Care Institute has done an excellent job reaching out to providers using a myriad of approaches: web-based seminars, a CME course, surveys of pediatricians, development of policy and survey papers, practical, practice improvement programs, and others. This broad approach has raised awareness of genetics in primary care for at least a segment of the provider population that is receptive to it. There is always more to be done, but this is a fantastic start, and addresses a real need in the primary care field.
>
> *—Melissa Parisi, MD, PhD, FAAP, FACMG, National Institute of Child Health and Development, National Institutes of Health*

Lessons Learned

Future initiatives targeting the integration of genetics into pediatric primary care should:

- continue to develop point-of-care resources for PCPs for genetic risk assessment and patient management
- explore solutions for the integration of family health history and genomics into EHRs
- frame genetics in primary care as valuable by coupling it with another pressing topic, such as pediatric EHRs, medical home, or obesity screening
- address provider reimbursement and payment related to genetic services

Translation of Evidence and Tools Into Practice

Quality improvement science can be used to implement a systems-level approach to integrating genetics into primary care. Clinical practices should partner with existing regional, state, and national entities and public health programs whenever possible. These partnerships can help patients and their families to better understand the role genetics plays in their lives and can enhance linkages between the medical home, families, and genetics professionals with important services and resources.

The following areas reflect gaps in evidence and implementation that should be considered for future practice-based initiatives:

- Implementation of regionally based quality improvement efforts (engage additional partners, such as Title V programs, payers, and healthcare institutions)
- Improvement of relationships between PCPs and genetics professionals
- Assessment of innovative approaches to accessing genetics professionals and genetic services
- Enhancement of EHRs and electronic tools to support family health history and practice-based quality improvement efforts
- Conducting research to assess patient outcomes for integrating genetics services into primary care
- Concertedly address transition to adult care for children with genetic conditions

Providing Continuing Medical Education to Healthcare Professionals

Educational material should be structured around how the information can be applied directly to clinical practice and around ensuring accessibility in multiple formats or vehicles, such as videos, reference guides, and case-based learning resources. Professional societies are encouraged to develop actionable recommendations for risk assessment, patient communication, and genetics referrals and evaluation. Educational initiatives should continue to develop condition-specific, population-level resources and to encourage widespread dissemination and adoption of tools. Maintenance of certification is a valuable and coveted benefit that can be used to engage PCPs, and efforts to offer this benefit should be considered when planning future educational initiatives.

Priorities for future initiatives include the following:

- Awareness of existing resources for genetics in primary care, particularly regarding genetic red flags, genetic testing, health supervision guidelines for patients with genetic conditions, and newborn screening
- Appropriate utilization of genetic tests and considerations for genetics consultations
- Practical strategies for incorporating systematic family health histories into health supervision visits, emphasizing the relative importance of comprehensive, targeted (just-in-time), and tailored family health histories for risk assessment and the accumulation of information over the pediatric period
- Providing case-based education that frames genetics as part of an integrated approach to primary care, as opposed to a siloed (and rare) component of medicine
- Ethical considerations of genetics and genomics
- Innovative modalities for increasing the competence of PCPs to discuss family health history and genetics with their patients, including videos of encounters between patients and providers

Workforce Training

Efforts related to training the future pediatric primary care workforce can be enhanced, evaluated, and promoted. The case-based modules for the continuity clinic that have been developed as part of the GPCI Resident Education Initiative have yet to be pilot tested in a real-life residency training program. The case series will undoubtedly benefit from feedback from program directors, continuity clinic preceptors, and residents.

Additionally, interactive, Web-based case modules based on the content included in the paper-based course series could be developed. How residency directors might adopt these resources and how they can be used most effectively in their programs should be considered. Moreover, the demonstrated success of these resources in pediatric residency programs may spill over to other healthcare professions and training programs, in particular family medicine, nursing, and physician assistant training programs. Genetic thinking must be integrated into the entire primary care workforce through training, formal education, and accountability. The model of the GPCI Resident Education Initiative seeks to embed genetic thinking within the current competencies and milestones, which would be appealing for other training programs that are faced with limitations of space in the curriculum and a lack of knowledgeable faculty to train and educate residents.

> **In years past we always thought of genetics as being very important to very few patients, now it's important to many, many patients. Rather than a few rare diseases that as pediatricians we've all learned something about and might see once in our practice lifetime, we're now going to be faced with the chronic common diseases and what are the genetic influences.**
>
> *—Tracy Trotter, MD, FAAP, National Coalition for Health Professional Education in Genetics*

Summary

GPCI was tasked with facilitating the translation of genetics information into resources that are relevant for the practice of primary care medicine. Over the 3-year cooperative agreement, the **GPCI has successfully established a foundation for the integration of genetics into pediatric primary care.** The GPCI has articulated what a pediatric PCP should know, and do, regarding genetic medicine in primary care. A plethora of educational and point-of-care resources have been developed, and the quality improvement project has demonstrated that incorporating genetics principles into a primary care setting is feasible, valued by patients and providers, and sustainable.

Dr Alan Guttmacher, MD, Director of the National Institute of Child Health and Human Development, explained to attendees of the 2013 Dive Into the Gene Pool conference that scientific advances about genetics and genomics in the growth and development in childhood, along with the origins of what has always thought to be adult onset disease, have significant implications for pediatric primary care clinicians. *"Primary care pediatrics in the relatively near future will really have the tools to be the gatekeeper to lifelong health. Anticipatory guidance—the bread-and-butter of pediatric practice—in the future won't just be anticipation of the next six days, weeks, or months, but rather the next six decades,"* said Dr Guttmacher.

Advances in genetics, genomics, pharmacogenomics, and other "genomic" sciences continue at a rapid rate and in order to stay abreast of ever-changing guidelines and tests, PCPs must have access to relevant information at the point of care. Existing resources should be promoted and marketing strategies should be aimed at making genetics seem less intimidating and within the accepted generalist roles. Payment models must support the integration of new genetic technologies into routine preventive and health supervision models of care. Professional societies should continue to develop specific guidelines, communicate messages to members that dispel existing myths regarding genetics, and provide direction regarding their respective roles in genetic medicine.

PCPs have varying levels of comfort regarding the application of genetics in primary care. Whether they chose to order genetic tests themselves, or refer their patients to genetic services, they need to be able to tier patient risk and be prepared to counsel their patients on the implications of test results. With the advancement of genetic technologies and continued workforce shortages of genetic professionals, PCPs will need to assume a greater responsibility towards genetic medicine for the anticipatory guidance and disease prevention that lie within their purview. The work of the GPCI is merely the beginning—and national partners and professional societies are encouraged to continue building upon this foundation.

As we learn more about genetics and genomics, the integration of those informational pieces into the routine practice of medicine is inescapable. Over the next several decades we will see PCPs called upon repeatedly to provide competent and effective interpretation of genetic information for patients and their parents.

—Barry Thompson, MD, FAAP, FACMG, American College of Medical Genetics and Genomics

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