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Committed to the attainment of optimal, physical, mental, and social health and well-being for all infants, children, adolescents, and young adults.

Genomic Medicine Working Group Meeting

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On behalf of the American Academy of Pediatrics

Chairperson, Committee on Genetics

Co-Chairperson, Genetics in Primary Care Institute

Initiatives in Genetics

- Committee on Genetics
- Section on Genetics and Birth Defects
- Division of Children with Special Needs
 - Genetics in Primary Care Institute
- Strategic Priority for Children's Health
 - Epigenetics Strategic Initiative

Genetics in Primary Care Institute

- A cooperative agreement between the American Academy of Pediatrics (AAP) and the Health Resources & Services Administration (HRSA)/Maternal & Child Health Bureau (MCHB), Genetic Services Branch
 - June 1, 2011 - May 31, 2014
- Vision of the Genetics in Primary Care Institute (GPCI) is to improve primary care provider (PCP) knowledge and provision of genetic medicine

Overview of the GPCI

- Goal 1
 - Utilize quality improvement (QI) science to develop a change package for the improved provision of genetic-related services
- Goal 2
 - Establish a technical assistance center to address systems and policy
- Goal 3
 - Embed the practice of genetic medicine into the future PCP workforce

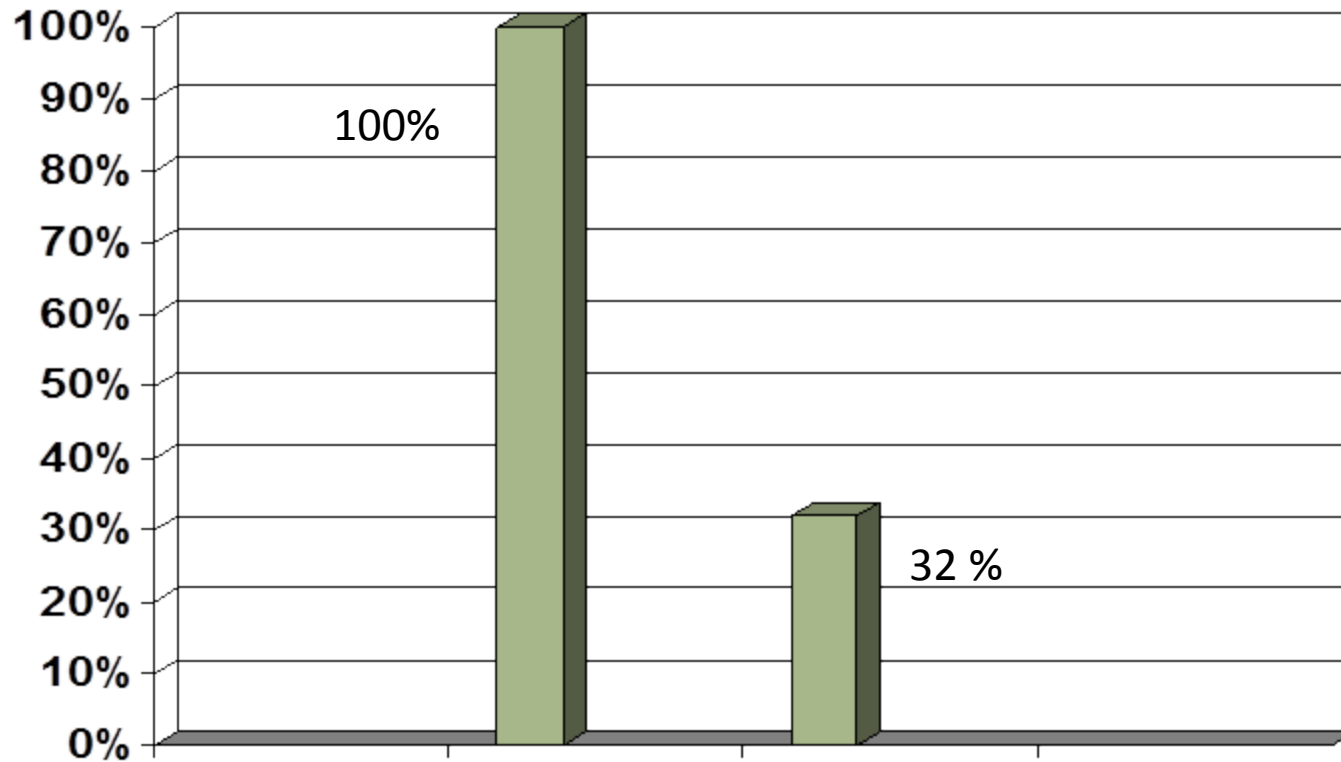
Needs Assessment of AAP Members

- Online survey of AAP members helped inform the QI project—February 2012 (88 respondents)
- Periodic Survey of AAP Fellows (approx. 1600 are surveyed) will be conducted in late 2013

Needs Assessment—Feb 2012

- Objective
 - Identify current practices and attitudes of pediatricians regarding genetic medicine
- Methods
 - 88 providers in the AAP QuIN membership (29% response rate)
 - Online, 43 item survey
 - Data analysis with Mann-Whitney two-sample statistic

Results—Attitudes

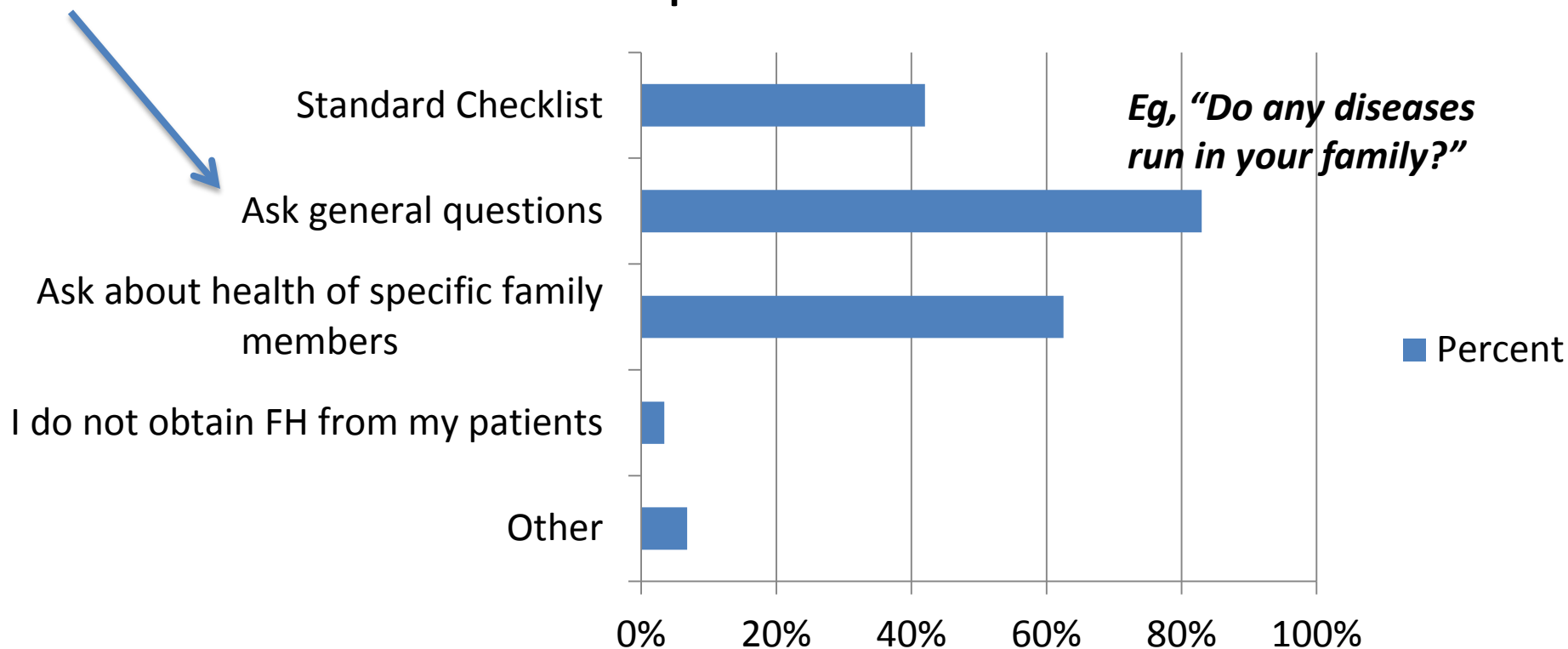


"Taking a family health history is important..."

"I gather a three generation family health history..." Strongly Agree/Agree

Results—Family History

“How do you usually collect a family health history from your patients”?



Results—Family History

- If they inquire about the health of family members, over 90% inquired about the health of siblings, parents, grandparents
 - Aunts/uncles—**55.3%**
 - Nieces/nephews—**23.5%**
 - Cousins—**29.4%**
 - Information least likely to be collected was age of family members, consanguinity, and ethnic background

Results—Current Practices

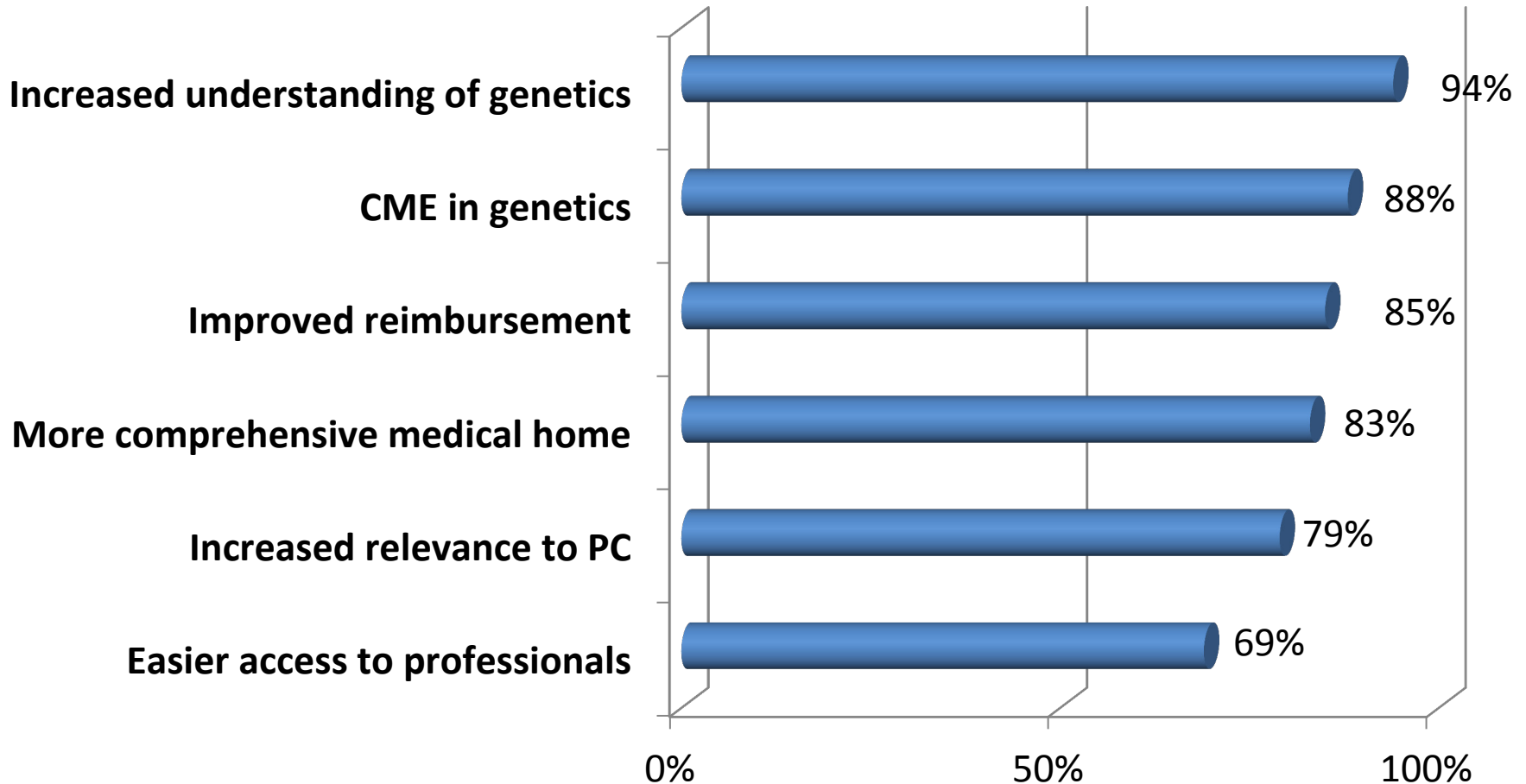
- 86% order genetic-based tests ≤ 3 annually
- 13% discuss with patients risks, benefits, and limitations of test in question
- Refer a mean of 4.8 patients a year to geneticist
 - 89% have access to a genetics professional
 - 75% have genetic professionals within 30 miles
- 83% report having a system for genetics referrals
- There was moderate to low awareness of national resources but overwhelmingly they had not been utilized or perceived useful
 - Out of 12 national genetics resources, less than 50% were aware of ACT Sheets or State Genetics Program, between 4% and 30% of respondents were familiar with the others

Results—Competency

- 49% agree or strongly agree that they feel competent in providing genetic medicine
 - Agreement was not associated with more recent training ($p=0.29$) or number of genetic tests ordered annually ($p=.84$)
- Of the 63 respondents that have an EHR, 65% report ability to easily & efficiently capture genetics information and FH as fair or poor

Results—Incentives

What would incentivize you to more effectively integrate genetic-based medicine into your practice?



Conclusions

- Convenience sample has slightly skewed results-- Sample group is more highly motivated and knowledgeable than providers nationally
- This sample reported few interactions with genetic patients, wide variability, expressed discomfort in GM
- Need for increased awareness, understanding, and access to education and resources

Stakeholder Interviews - Methods

- Objective
 - Identify and understand current barriers and means to address barriers in genetics in primary care
- Methods
 - Conducted 7 in-depth interviews with key project stakeholders and experts
 - Interviewees were identified by the AAP
 - Interviews were conducted via phone and lasted 45-60 minutes

Stakeholder Interview Topics

- Interviews covered the following topic areas:
 - evolution of genetics in primary care
 - barriers to incorporating genetics into primary care
 - key competencies needed by primary care providers (PCPs) to incorporate genetics into practice
 - moving genetics in primary care forward
 - actionable items for the Genetics in Primary Care Institute (GPCI)

Stakeholder Interviews - Barriers

- PCPs lack knowledge and comfort with genetics

“There is a lack of fundamental knowledge about what things mean and what to do, and people are uncertain of where to turn to for specialty help.”

- No time to incorporate into practice
- Lack of education and training in genetics

“Residents are stretched very thin on what they have to learn, and genetics competes with other areas residents have to learn.”

Stakeholder Interviews - Barriers

- Low accessibility of genetic counselors and specialists
- Lack of involvement of PCPs in genetic projects and initiatives
- Lack of knowledge of what is reimbursed

“I don’t know which tests covered and which ones are not. Some of these tests run into the hundreds, even thousands of dollars and I don’t want to order something that a family is going to have to pay for... if I send them to a specialist they know which ones are covered and which ones are not.”

Stakeholder Interviews – Moving Genetics in Primary Care Forward

- Increased provider education and training (e.g., formal classes, training in medical school and residency, modeling behavior, connecting with high interest areas)
- Evidence to support genetics in primary care is important and impacts outcomes
- Additional ideas included:
 - Better ways to collect and store family history
 - Improved care coordination plans between PCPs and specialist
 - Increase/including genetics information on board exams
 - Incorporating genetics into continuing medical education and professional meetings

Impact of Low Genetic Literacy

- Management of diagnostic workup of positive NBS
 - More than half of pediatricians prefer not to manage
- Advanced molecular genetic testing expand ability to diagnose and treat genetic conditions
 - Providers not aware of their role in testing or in treatment advancements

Barriers

- **Scope unclear**
- **Boundaries unclear**
- **How to order testing**
- **“State-of-the-art” confusion**
- **Extended family**
- **Prevention context**
- **Test and terminology interpretation**
- **Natural history**
- **Consultations**
- **Monitoring**

Genetic Literacy in Primary Care Colloquium

- Purpose and Overview
 - Increase the knowledge base about, and awareness of, genetic literacy in the medical home
 - October 2-3, 2012
 - Develop and present papers, formulate recommendations
- Outcomes
 - Overarching consensus statement will accompany papers published as a supplement to *Pediatrics* (early 2013)

Colloquium Structure

Focus Areas

- Family History
- Genomics
- Genetic Literacy
- Epigenetics
- Primary Care and Genetics

Guests

- AAFP
- AAP
- ACGME
- ACMG
- APA
- CDC
- Genetic Alliance
- HRSA
- NAPNAP
- NCHPEG
- NIH

Colloquium Consensus Statement

1. **Define how pediatric primary care providers should use genetics and genomics in practice.**
 - a. Recognize that many primary care providers are already using 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.

Colloquium Consensus Statement

2. **Define, develop, and provide the tools/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/family education and support tools that are culturally sensitive and literacy and language appropriate.
 - e. Facilitate access to appropriate family support and advocacy.

Colloquium Consensus Statement

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 pre-professional to postgraduate education.

Colloquium Consensus Statement

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 or needed infrastructure to facilitate the research agenda.

AAP Plans to Address Shortfalls

- Genetics in Primary Care Institute
 - Technical Assistance Center
(www.geneticsinprimarycare.org)
 - Educational Webinars
 - Promotion of vetted tools and strategies
 - Development of Family History Tool for Pediatric Providers
 - Residency training initiatives

AAP Plans to Address Shortfalls

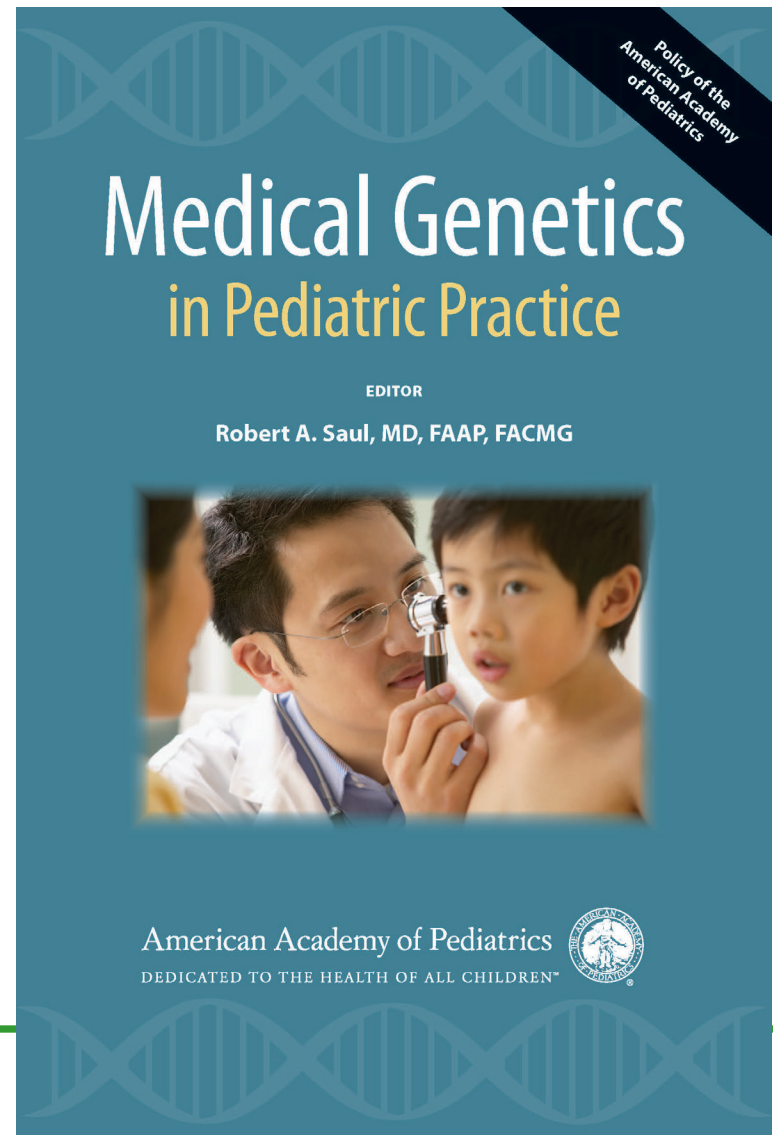
- Genetics in Primary Care Institute
 - Educational Webinars
 - Series of 10, one-half hour talks in 2012
 - Well-received (audience and reviews)
 - Archived on website
(<http://www.medicalhomeinfo.org/GPCI.aspx#webinar>)
 - Organization from the AAP's Genetics Handbook
 - Template for collaborative efforts for professional societies and educational efforts

AAP Plans to Address Shortfalls

- AAP's Newest Strategic Planning Priority
 - “Genetics, Genomics, and Epigenetics”
- CME Conference (Funded in part by Centers for Disease Control and Prevention—NCBDDDD)
 - “Dive Into the Gene Pool -- *Integrating genetics and genomics into your pediatric primary care practice*”
 - Two-day conference planned for August 2013 for a public audience of pediatric PCP

AAP Plans to Address Shortfalls

- Committee on Genetics
 - New AAP Manual:
Medical Genetics in Pediatric Practice
 - 23 Chapters
 - Over 400 pages
 - Available Spring 2013
 - Companion mobile app under development



AAP Plans to Address Shortfalls

- Committee on Genetics
 - Policy Statements
(Recently Published)
 - Health Supervision for Children with Down Syndrome
 - Health Supervision for Children with Fragile X Syndrome
 - Health Supervision for Children with Prader Willi Syndrome

AAP Plans to Address Shortfalls

- Committee on Genetics
 - Policy Statements
(Selected Titles Under Development)
 - Ethical Issues with Genetic Testing in Pediatric Practice
 - Clinical Genetic Evaluation of the Child with Intellectual Disability or Developmental Delay
 - Health Supervision for Children with Marfan Syndrome
 - Prenatal Screening and Diagnosis for Pediatricians

AAP Plans to Address Shortfalls

- Section on Genetics and Birth Defects
 - Educational programming at AAP's Annual National Conference & Exhibition.
 - Articles on genetics-related topics in AAP's monthly member news magazine **AAP News**.
 - Supports genetics-related content in **AAP Grand Rounds** and other Academy publications.
 - SOGBD-member-only online open forum discussion group.

Questions?

