Integrating Genetics into Primary Care: Why Does It Matter?

Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute
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Tuesday, March 20, 2012
12:00-12:30pm Central

Learning Objectives

1. Recognize how advances in genetics and genomics can contribute to pediatric primary care.

2. Describe the importance of care coordination within the medical home for children with genetic conditions.

Role of Primary Care

- PCPs on the frontline of care
  - Shortage of clinical geneticists
  - PCPs need fundamental knowledge base in genetic/genomic medicine
- Family health history
- Genetic screening
  - Newborn Screening (NBS)
  - Adult onset disease
  - Direct-to-consumer testing
PCPs Role in Genetic Screening

- Educate
- Explain
- Coordinate care with subspecialist
  - Refer
  - Initiate treatment
  - Coordinate diagnostic testing
  - Assure appropriate follow-up
- Counsel

Pediatric Primary Care Provider Assessment Perceived Competencies:

100% in survey agreed that health-related genetic information has important social, emotional, and psychological implications for individuals and families.

Source: GPCI Needs Assessment with Quality Improvement and Innovation Network (QuIIN) Survey, 2012

Pediatric Primary Care Provider Assessment Perceived Competencies:

“I feel competent in providing health care to my patients that is related to genetics and genomics.”

48.9% Disagree
1.1% Strongly Disagree
42.2% Agree
6.8% Strongly Agree

Source: GPCI Needs Assessment with Quality Improvement and Innovation Network (QuIIN) Survey, 2012
"Taking a family health history is important..."

"I gather a three generation family health history..."

Source: GPCI Needs Assessment with Quality Improvement and Innovation Network (QuIIN) Survey, 2012

Pediatric Primary Care Provider Assessment Perceived Competencies:

**Family Health History**

| Percentage | 100% | 32% |

"The health care (conversations and assessments) that I provide to my patients would be enhanced by professional educational opportunities (CME, lectures) related to genetics and genomics."

- **Strongly Disagree**
- **Disagree**
- **Agree**
- **Strongly Agree**

Source: GPCI Needs Assessment with Quality Improvement and Innovation Network (QuIIN) Survey, 2012
Case Study

- 20 m/o former 26 week pre-term female without chronic lung disease
- Started having recurrent bouts of wheezing
- Prenatal screening - mother positive for common CF mutation; father’s screen negative
- Sweat Chloride Test - 30 LA, 31 RA (0-40)
- Pulmonologist impression - viral induced asthma, rule-out CF variant
- DNA testing: negative for CF

Impact of Genetics/Genomics on Primary Care

Infectious disease          Genetics/Genomics

Acute Care Model
- Diagnosis
- Treatment

Chronic Care Model
- Lifelong implications
- Requires chronic condition management
- Family-Centered Medical Home

Medical Home for Hereditable Conditions
New Approaches in Genetics

Prevention

Diagnosis

Treatment

Prevention: Newborn Screening

Newborn Screening
Diagnosis:
The First Genomic Test

Array Comparative Genomic Hybridization

- Reference DNA
- Test DNA
- Mix
- Block repeated sequences
- Hybridize
- Microarray with oligonucleotides
Cumulative Pace of Disease Gene Discovery 1981-2005

Medullary Thyroid Carcinoma
Family History

https://familyhistory.hhs.gov/fhh-web/home.action

Cost per Genome

Exome vs Genome Sequencing
The Diagnostic Odyssey

Treatment: Lysosomal Storage Disorders

- Enzyme Replacement Therapy
  - Gaucher disease
  - Fabry disease
  - Pompe disease
  - Mucopolysaccharidoses
  - Others in pipeline

Small Molecule Therapies: NF1

- Plexiform Neurofibroma
- Learning Disability
- Low-grade Glioma
- Malignant Peripheral Nerve Sheath Tumor
- STOPN
- STARS
- RAD001
- MPNST
Pharmacogenetics: Thiopurine S-methyltransferase

- S-methylation of heterocyclic sulfhydryls
- 6-mercaptopurine, 6-thioguanine
- Leukemia, autoimmune diseases
- TPMT involved in inactivation of drugs
- 10% subjects have intermediate activity, 0.3% low activity
- Low or intermediate activity associated with increased toxicity and requires lowering of dose

Perceptions About Genetics

- Genetics deals only with rare disorders
  *Both rare and common disorders can have a genetic basis*
- Children should not be tested for genetic disorders
  *Only applies to adult-onset disorders*
- Insurance doesn’t pay for genetic testing
  *Most insurance companies pay for diagnostic testing*
- Genetic testing leads to discrimination
  *GINA bill/State laws*
- Genetic disorders are not treatable
  *Many are, and many more soon will be*

The Pediatrician’s Role

- Clinical Suspicion
- First Response to Newborn Screen
- Referral to Specialist
- Support
- Ongoing Management
- Medical Home
Genetics in Primary Care Institute

• 3 year cooperative agreement
  • HRSA/MCHB and AAP
• Goals
  • Develop strategies to enhance PCP knowledge regarding genetic-related services
  • Establish a technical assistance center
  • Address residency training needs

GPCI Priority Areas

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

Questions?

Thank you for your participation!

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www.medicalhomeinfo.org/gpci
Time Out for Genetics

Upcoming Webinar
Building an Accurate Family History, Constructing a Pedigree – An Overview for Primary Care

Thursday, April 26
12:00-12:30pm Central

Register:
https://www2.gotomeeting.com/register/486497370