Genetics in Primary Care: Why Does it Matter

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March 8, 2013
Objective

• To Understand the importance of genetics in primary care, through practical application and parent experience.
Disclosures

• “I have no relevant financial relationships with the manufacturers(s) of any commercial products(s) and/or provider of commercial services discussed in this CME activity.”

• I do not intend to discuss an unapproved/investigative use of a commercial product/device in my presentation.
Role of Primary Care

- PCPs on the frontline of care
  - Acute care
  - Preventative Care
  - Chronic Care

- Family health history

- Screening and Surveillance
  - Growth and Development
  - Newborn Screening (NBS)
  - Genetic Screening
Newborn Screening

• Began in the 1960s with PKU
• Rapid expansion in the number of congenital conditions
• Genetic testing on virtually every newborn in the last 5 decades
• Public health model for early diagnosis and treatment of genetic diseases
• Provide both short-term and long-term follow-up for those patients
PCPs Role in Genetic Screening

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

• Counsel
• Provide Long-term care
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
GPCI QuIIN Needs Assessment

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

- 48.9% Disagree
- 43.2% Agree
- 6.8% Strongly Agree
- 1.1% Strongly Disagree
“Taking a family health history is important…”

“I gather a three generation family health history…”
GPCI QuIN Needs Assessment

Genetic Testing

- Weekly: 1.2%
- Monthly: 14%
- 2-3/year: 46.5%
- 0-1/year: 34.9%
- Never: 3.5%
“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: 2.3%
- Disagree: 34.9%
- Agree: 62.8%
- Strongly Agree: 34.9%
Perceived Barriers

• Time pressures to comply with practice guidelines

• Lack knowledge, training, experience, and confidence
  – understanding of basic genetics
  – collection and interpretation of family health histories
  – Ordering, interpreting, and acting on genetic tests.

• Genetics deals only with rare disorders

• Insurance doesn't pay for genetic testing

• Genetic testing leads to discrimination

• Genetic disorders are not treatable
# Genomic Advances

<table>
<thead>
<tr>
<th>Traditionally:</th>
<th>Now:</th>
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<tbody>
<tr>
<td>• Genetic issues - rare occurrences</td>
<td>• Increased public awareness</td>
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<tr>
<td>• Large role in the health care of few</td>
<td>• Increased demand for advice</td>
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<tr>
<td>• Small role in the health of many</td>
<td>• Increased availability of DNA-based testing</td>
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<td>• Integration of “genomic thinking”</td>
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<td>• Shortage of clinical geneticists</td>
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Human Genome Project

- Exponential growth in genetic knowledge,
- Continually expanding array of genetic tests.
- The public health burden
- Evaluation of clinical condition or family history
Pharmacogentics

• Tests for gene variants in drug-metabolizing enzymes
• Predict individual response
• Allow personalized drug prescribing
• Reduce adverse reactions
• Increase the efficacy of drug treatment
The Ethical, Legal, and Social Issues of Genomics in Primary Care Practice

- Confidentiality
- genetic discrimination
- direct-to-consumer genetic testing
- predictive genetic testing in children
- duty to contact
- access
- health disparities
- cultural awareness
- race and genetics
The Lifespan

• Monitor genetic, epigenetic and environmental factors
• Obtain and update the family history,
• Constant re-evaluation of past diagnosis or non-diagnosis
• Incorporate new genomic applications
• Inform adult conditions whose origins may be in early life events
Competencies

• Replace the concept of genetic issues being rare occurrences
• Integrate “genetic thinking” into every encounter
  – prevention
  – assessment,
  – evaluation and diagnosis,
  – ordering and interpreting genetic testing,
  – communication with families,
  – appropriate referrals,
  – management or co-management.
Impact of Genetics/Genomics on Primary Care

Infectious Disease

- Acute Care Model
  - Diagnosis
  - Treatment

Genetics/Genomics

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

- Care Coordination
- Co-management
- Community Based Resources
- Screening and Surveillance
- Quality Improvement
- Transitions

Medical Home Family

Practice Improvement Network
from the Quality Improvement Innovation Networks of the American Academy of Pediatrics

Genetics in Primary Care Institute
Diagnostic Odyssey

- Inform treatment decisions
- Early supportive intervention for the child and family
- Prognosis
- Aid reproductive decision-making,
- Identify at-risk family members
- Facilitate clinical research studies may be available to family
- Alleviate the psychosocial burden families.
Andy

- 22q11.2 Deletion
- Velocardiofacial Syndrome
- Expressive Language Disorder
- Growth Hormone Deficiency
- Attention Deficit Disorder
- Chronic Tympanic Membrane Perforations
Summary

- Identify at risk individuals
- Recognize genetic conditions
- Monitor health
- Provide information
- Provide a medical home
- Recognize family issues
- Knowledge of genetics services
- Appropriate referrals
- Facilitate services.