Heard About Genetic Counseling?
What Does it Mean for You, Patients, and Families?

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Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute
Presenter

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  – Certified and licensed genetic counselor
  – Assistant Professor in the Department of Pediatrics at the University of Utah
  – Immediate past president of the National Society of Genetic Counselors (NSGC)
Ms. Dent has no financial relationships or conflicts of interest to disclose relevant to this presentation.
Learning Objectives

At the end of this presentation, participants should be able to:

1. Define genetic counseling
2. Describe the role of a genetic counselor
3. List three ways patients and families can benefit from meeting with a genetic counselor
4. Find a genetic counselor in your area
What is Genetic Counseling?

- The process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates:
  - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
  - Education about inheritance, testing management, prevention, resources and research
  - Counseling to promote informed choices and adaptation to the risk or condition

National Society of Genetic Counselors, 2005
Purpose of Genetic Counseling

• Educate patient about the way heredity contributes to genetic disease:
  – Specific condition and pattern of inheritance
  – His/her individual risk of developing genetic disease

• Help patient understand options for dealing with increased risk
• Provide guidance about what diagnosis means
• Provide psychosocial support
• Aid patient in choosing a course of action that is personally appropriate

Bernhardt BA et al., AJMG 2000; 94: 189-197.
Who Provides Genetic Counseling?

• Healthcare providers
  – Physicians
    • OB/GYN, pediatricians, family practice, internal medicine, oncologists, neurologists, medical geneticists, etc.
  – Physician Assistants
  – Nurses
  – Genetic Counselors
What is a Genetic Counselor?

- Health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling

- Members of a healthcare team who provide:
  - Information and support to families of patients with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions
What is the Role of a Genetic Counselor?

• Serve as patient advocates
• Serve as educators and resources for other health care professionals
• Engage in research activities
Skills of Genetic Counselors

• Deep and broad knowledge of genetics
• Ability to tailor, translate and communicate complex information in a clear, relevant way for a broad range of audiences
• Strong interpersonal skills, emotional intelligence, and self-awareness
Skills of Genetic Counselors

- Ability to dissect and analyze a complex problem
- Research skills
- In-depth knowledge of healthcare delivery
Why is this Important?

- Impact of genetic conditions on public health

Approximate Prevalence of Genetic Disease in the General Population*

<table>
<thead>
<tr>
<th>Type of Genetic Disease</th>
<th>Lifetime prevalence per 1000 persons</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autosomal dominant</td>
<td>3 to 9.5</td>
</tr>
<tr>
<td>Autosomal recessive</td>
<td>2 to 2.5</td>
</tr>
<tr>
<td>X-linked</td>
<td>0.5 to 2</td>
</tr>
<tr>
<td>Chromosome disorder</td>
<td>6 to 9</td>
</tr>
<tr>
<td>Congenital malformation</td>
<td>20 to 50</td>
</tr>
<tr>
<td><strong>Total:</strong></td>
<td><strong>31.5 to 73</strong></td>
</tr>
</tbody>
</table>

- Patients with genetic conditions constitute an important portion of the world population with special health care needs

Impact of Genetic Diagnosis

• Permanent, chronic, often no cure
• Long-term issues regarding independence, care
• Sense of responsibility/guilt
• Age dependent/life stages
• Severity of disorder
• Insurability and employability issues
• Stigma
GCs: Partners in Practice

• Genetic professionals can be your partner in ensuring that your patients benefit from advances in genetics.
  – Help HCPs and patients bridge the gap between science and care
  – Tailor complex info to patient’s understanding
  – Assemble and interpret complex genetic information
    • Case mgmt, appropriate tests, risk assessment, targeted family history, etc.
  – Trained in resolving ethical dilemmas
Top Specialty Areas of GCs

Figure 5. Top Specialty Areas

- Prenatal: 29%
- Cancer: 25%
- Pediatric: 13%
- Other*: 33%

*Other includes areas like prenatal, cancer, and pediatric.
Other Specialties

- Laboratory
- Molecular / cytogenetics / biochemical testing
- Metabolic disease (LSDs)
- Administration
- Education; public and professional
- Cardiology
- General genetics
- Research
- Neurogenetics
- Genetic testing
- Adult screening
- Genomic medicine
- Infertility

- ART/IVF
- Personal genomics/genomic profiling
- PGD/preconception
- Public health
- Population based/biobanking
- Newborn screening
- Hematology
- ELSI
- Psychiatry
- Teratogens
- Pharmacogenetics
- Public policy
- Minority health
- Support group/advocacy
Where do you find GCs?

**Figure 4. Primary Work Setting**

- University Medical Center: 36%
- Private Hospital/Medical Facility: 17%
- Other*: 13%
- Public Hospital/Medical Facility: 17%
- Physician’s Private Practice: 5%
- Health Maintenance Organization: 3%
- Diagnostic Laboratory - Commercial: 9%

*Other* includes a variety of settings such as non-profit organizations, independent genetic counseling practices, and other private practices.
Primary roles of GCs

- 77% provide direct patient care
- 15% work in non-clinical roles
  - Laboratory support
  - Management
  - Sales/marketing/business development
  - Project management
  - Teaching/education/supervising students
  - Customer liaison
  - Clinical coordination/case management

Components of a GC Interaction

• Information gathering
  – Contract with patient (review reason for appt)
  – Records review
  – Medical and family history

• Establishing or verifying a diagnosis
  – History
  – Physical exam (not by GC)

• Risk assessment
  – Pedigree
  – Recurrence risk of known condition
  – Empiric recurrence risk
  – Testing
Components of a GC Interaction

• Information giving
  – Discussion of natural history of a diagnosis
  – Decision making

• Psychosocial assessment and counseling
  – On-going client support

• Follow up
  – Support resources
Psychosocial issues

• Grief reactions: denial, fear, sadness, anger, loneliness
• Readjustment of expectations
• Potential family and marital strain
• Impact on siblings/unaffected family
• What to tell other family members, friends, teachers?
• Stigma of the terminology: ‘mental retardation/intellectual disability’
Case example 1: Pediatrics

- New diagnosis of Angelman syndrome due to maternal deletion of 15q11.2
Case 1: Role of Genetic Counselor

• Role of genetic counselor:
  – Natural hx of AS
  – Anticipatory guidance for AS
  – Explain inheritance
  – Recurrence risks
  – Psychosocial support
  – Resources
  – Follow up
  – Options for future pregnancies
Case example 2: Prenatal

• A pregnant woman has a brother with cystic fibrosis and wants to know the chance her baby could also have the condition.
Case 2: Role of Genetic Counselor

• Role of genetic counselor:
  – Known mutations in family?
  – Father’s genetic status?
  – Inheritance & recurrence risks
  – Arrange testing (if desired)
  – Psychosocial support
  – Follow up
  – Options for future pregnancies
Case example 3: Cancer

- A 30 year old woman has a family history of breast cancer and wants to know her risk of also developing breast cancer
Case 3: Role of Genetic Counselor

- Role of genetic counselor:
  - Evaluate family history
  - Testing options?
  - Anticipatory guidance
  - Natural hx of condition
  - Treatment options / management
  - Recurrence risks
  - Psychosocial support / aid in decision-making
  - Resources
Genetic Testing

• Should always be performed in context of genetic counseling
  – Discuss medical and social concerns
  – Arrange appropriate testing
  – Review implications of testing
  – How use the information?
  – Provide psychosocial support
Resources for you and your patients

www.nsgc.org
Resources for you and your patients

- American Board of Genetic Counseling
  www.abgc.net

- March of Dimes: Genetics and Your Practice
  www.marchofdimes.com/gyponline/index.bm2
Questions?
Thank you for your participation!

For more information, please contact
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www.medicalhomeinfo.org/GPCI.aspx
Time Out for Genetics

Registration is now open for

“Genetics and Coding: What the Primary Care Provider Needs to Know”

Thursday, September 27
12:00 - 12:30pm Central

https://www2.gotomeeting.com/register/414021762