Genetic Counseling in Primary Care

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1pm

Integrating Genetics into Your Practice Webinar Series
Presented by the Genetics in Primary Care Institute
Faculty

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  - Associate Director, Clinical & Continuing Education, The Jackson Laboratory
  - Formerly at NCHPEG
Ms Edelman has no financial relationships or conflicts of interest to disclose relevant to this presentation.
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Learning Objectives

• Recognize key competencies and skills in the practice of genetic counseling.
• Apply genetic counseling skills to the risk assessment, evaluation, and management of patients in primary care clinic.
• Explore opportunities for collaboration with genetic counselors for improved patient care.
Utilizing Genetic Counseling

Risk Assessment

Consult

Communicate/Counsel
Case Scenario: Sara

Sara

• 10 month healthy female presenting for well child visit
• Family history of Duchenne Muscular Dystrophy (DMD)
Case Scenario: Sara

Tyler 4 y DMD

Sara 10 mo
Case Scenario: Sara

• Will Sara develop DMD?
• What is the chance to have another child with DMD?
• What is the risk to relatives?
Case Scenario: Sara

PCP risk assessment

☑ Known genetic syndrome
☑ DMD is X-linked
☑ Sara may be a carrier
☑ Sara may develop symptoms
☑ Sara’s parents have an increased chance to have another child with DMD

➔ Family appropriate for genetic counseling
Case Scenario: Sara

Q. How do you address the family concerns?

- Offer DMD testing for Sara
- Discuss inheritance and recurrence risks
- Refer back to neurology
- Refer to genetic counseling
Case Scenario: Sara

A. Refer or consult with genetics expert; provide anticipatory guidance

- Offer DMD testing for Sara
- Discuss inheritance and recurrence risks
- Refer back to neurology
- Refer to genetic counseling
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
Slide courtesy of K. Dent
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.
Slide courtesy of K. Dent
Who Provides Genetic Counseling?

• Healthcare providers
  – Physicians
    • Medical geneticists, OB/GYN, pediatricians, family practice, internal medicine, oncologists, neurologists, etc
  – Physician Assistants
  – Advanced Practice Nurses
  – 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
Where do you find GCs?

**Figure 4. Primary Work Setting**

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

*Includes Genetic Counselors employed in non-clinical settings.
Geneticist vs. Genetic Counselor

• Team approach
• GCs > Medical Geneticists
• Family may see one or both professionals
• Both
  – Perform risk assessment
  – Consider a differential and evaluation plan
  – Order and interpret genetic testing
Accessing Genetic Counseling Services

• Within your institution
• National Society of Genetic Counselors (www.nsgc.org)
• American College of Medical Genetics (www.acmg.net)
• Telemedicine
• Regional Genetics Collaborative (www.nccrcg.org)
Finding a Genetics Professional

Experience the value of a genetic counselor. Increase patient satisfaction and enhance the services you provide to your patients.

The role of genetic testing in healthcare grows more prevalent and complex everyday. Do you have the best resources to provide the most comprehensive, personalized care for your patients?

Healthcare Providers

How can a genetic counselor help my practice?

Genetic counselors can help you incorporate the latest in genetic science into your practice of care by providing comprehensive genetic and genomic services to your patients.

Learn More >>

Your Genetic Health

How can genetic counselors help you and your family?

Genetic counselors can give you the personalized help you need when it comes to you and your family’s genetic health.

Learn More >>

Prospective Students

Interested in pursuing a career in genetic counseling?

NSGC is your resource for pursuing or advancing your career as a genetic counselor.

Learn More >>

Find a Genetic Counselor

Interested in contacting a genetic counselor in your area?

Find a Genetic Counselor
Finding a Genetics Professional

FIND A GENETIC COUNSELOR

NSGC's Find a Genetic Counselor directory has been developed to assist physicians, patients and other genetic counselors in locating genetic counseling services and for students curious about the profession. Genetic Counselors can be searched by State, City, Counselor's Name, Institution, Work Setting, Type of Specialty or Zip Code. We hope you find this service helpful.

Additional Resources and Information

Information on Services of a Genetic Counselor

- Different patient services that each counselor serves are listed under the counselor's name. Definition of services.
- Most counselors will perform a brief intake to determine whether or not an appointment for a genetic consultation would be of benefit to you. Longer consultations by phone are usually not possible.
- A scheduled appointment would be necessary to discuss your concerns.
- Each center varies with regard to referral requirements, payment methods, and services.

Disclaimer of Warranties and Liabilities

The NSGC Find A Counselor directory is not a referral service but rather a service to provide an up-to-date list of NSGC members. The NSGC makes no representation or warranty about the professional skills of those genetic counselors listed in the directory, nor does the NSGC accept or assume any responsibility for the services they provide.

Search for a Genetic Counselor:

Search For:  ○ Any criteria  ○ Exact criteria

State/Province:  Any State/Province

OR

Near (Postal Code or City, State):  21224

Within:  25 miles

First Name:  

Last Name:  

Types of Specialization:  

Pediatric

POD/Preconception

Personalized Genomic Medicine

Prenatal

Student Contact Welcome:  

Sees Patients:  

Telephone Counseling Service:  

Board Certified:  

Search
Finding a Genetics Professional
Finding a Genetics Professional

**Search for a Genetic Counselor:**

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<tr>
<td>Meghan Dumas</td>
<td>Maine Medical Partners Pediatric Specialty Care 1577 Congress St 2nd Floor Portland, ME, US 4102</td>
<td>(207) 862-5522 (phone) EMAIL</td>
</tr>
<tr>
<td>Sara A. Ellingwood, MS</td>
<td>Maine Medical Partners Pediatric Specialty Care 1577 Congress St Ste 2 Portland, ME, US 04102-2169</td>
<td>(207) 862-5522 (phone) (207) 774-1814 (fax) EMAIL</td>
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- Sees Patients
- Student Contact Welcome
- Pediatric
- Board Certified
Telemedicine Genetic Counseling

Telegenetics: a systematic review of telemedicine in genetics services

Jennifer S. Hilgart, BSc, MSc¹, Julie A. Hayward, BSc¹, Bernadette Coles, MSc² and Rachel Iredale, MA, PhD³

Purpose: Telemedicine is being increasingly used in many areas of health care, particularly to reduce the barriers that rural populations face in accessing health-care services. Telemedicine may also be effectively utilized in clinical genetics services—an application that has been termed “telegenetics.”

Methods: A systematic review of the literature was conducted to identify studies of genetic consultations carried out through video-conferencing so as to determine whether conclusions can be drawn about the value of telegenetics. A total of 14 articles reporting data from 12 separate studies met the inclusion criteria.

Results: In a majority of these studies, patients received their telegenetics consultation at a local clinic or outreach center, from where they communicated via a synchronous video link with a genetics with telegenetics, and patients were generally more receptive to telegenetics than the genetics practitioners were. The studies had limitations of small sample sizes and lack of statistical analyses.

Conclusions: This review suggests that telegenetics may be a useful tool for providing routine counseling and has the potential to evaluate pediatric patients with suspected genetic conditions. Prospective, fully powered studies of telegenetics that explore the accuracy of diagnoses and patient outcomes are needed to allow informed decisions to be made about the appropriate use of telemedicine in genetics service delivery.

Genet Med 2012;14(9):765–776

Keywords: clinical genetics; genetic counseling; systematic review;
Telemedicine Genetic Counseling
Case Scenario: Sara

• Referral to genetics for DMD genetic counseling
• Genetic counselor meets with family
  • Educates family about condition and pattern of inheritance
  • Counsels family about risks
  • Discusses risks, benefits, and limitations of testing options
  • Provides psychosocial counseling
  • Aids family in choosing a course of action
  • Communicates plan to PCP
Genetic Counseling for DMD risks

• Inheritance of DMD
• De novo vs. inherited gene change
• Carrier testing for Sara’s mother
• Results affecting Sara and future siblings risks
• Counseling about natural history of DMD in boys and carrier girls
Genetic Counseling for Carrier Testing

- Risks, benefits, and limitations of DMD carrier testing
- Potential outcomes of testing
- Logistics – timeline, insurance coverage
- Facilitates patient decision making
- Shared plan
Genetic Results may Impact Families Differently

- Guilt & Shame
- Privacy
- Relief from uncertainty
- Stigma & Discrimination
Summary of Scenario

• Role of GC
  – Natural history of DMD
  – Explain inheritance
  – Recurrence risks
  – Psychosocial support / aid in decision-making
  – Coordinate genetic testing
  – Resources
  – Options for future pregnancies

www.nchpeg.org/neuro/products/counseling-family-recurrence-risk
Case Scenario: Marcus

- 45 y/o new patient
- No significant health problems
- Family history of cancer
Case Scenario: Marcus

Breast cancer

Lung ca smoker

45

14 12
Case Scenario: Marcus

Q. What is your next step?

- Refer to genetic counseling
- Collect more information about the family
- Order genetic testing for BRCA1/2
Case Scenario: Marcus

- Genetic counseling skills
  - Collect comprehensive family history
Case Scenario: Marcus

• Genetic counseling skills
  ✓ Risk assessment
  • Collect comprehensive family history
  • Identify red flags: multiple relatives with condition, early-onset disease, bilateral disease, possible dominant pattern
  • Analysis: Increased risk for hereditary breast and ovarian cancer
  • Communicate with family about assessment
Case Scenario: Marcus

Risk Assessment

✓ Increased risk for inherited breast cancer
Case Scenario: Marcus

Risk Assessment

✓ Increased risk for inherited breast cancer

Q. What is your next step?

☐ Refer to genetic counseling
☐ Counsel about hereditary breast cancer
☐ Order genetic testing for BRCA1/2
Case Scenario: Marcus

- Genetic counseling skills
  - Evaluation & testing decisions
    - Create and narrow differential = HBOC
    - Develop strategy for cost effective testing
    - Identify most likely variants and choose testing

[Genetic counseling tree diagram]

- Lung cancer smoker
- d. 28 MVA
- dx 30s
- dx mid-40s
- dx 30s
- 14
- 12
Case Scenario: Marcus

• Genetic counseling skills
  ✓ Evaluation and testing decisions
  ✓ Pre-test counseling and informed consent
  ✓ Coordinates testing
    • Pre-authorization, if needed

Lung ca smoker

d. 28 MVA

45

14
12

dx 30s
dx mid-40s

dx 30s
Case Scenario: Marcus

- Lung cancer smoker
- d. 28 MVA
- dx 30s
- dx mid-40s
- BRCA2+
- dx 30s
Case Scenario: Marcus

Lung ca smoker

d. 28 MVA

45

BRCA2+

dx 30s

dx mid-40s

BRCA2+

dx 30s

14

12
Case Scenario: Marcus

Genetic Testing & Interpretation

✓ Familial BRCA2 mutation

Q. What is your next step?

☐ Refer to genetic counseling
☐ Counsel Marcus about his cancer risks
☐ Order genetic testing for BRCA1/2 for Marcus’s daughters
Case Scenario: Marcus

• Genetic counseling skills
  ✓ Genetic testing and interpretation
    • Interpret result in context of patient/family history
    • Reference guidelines and best practice
    • Counsels family about result, cancer risks, and general management options
    • Provides psychosocial counseling
    • Coordinates referrals/care with PCP and specialists
Case Scenario: Marcus

• Genetic counseling skills
  ✓ Genetic testing and interpretation
    • Interpret result in context of patient/family history
    • Reference guidelines and best practice
    • Counsels family about result, cancer risks, and general management options
    • Provides psychosocial counseling
    • Coordinates referrals/care with PCP and specialists
      • Breast: Clinical breast exams and mammography
      • Prostate: DRE & PSA
Case Scenario: Marcus

- Genetic counseling skills
  - Genetic testing and interpretation
  - Genetic testing of minors
    - Reference guidelines and best practice
    - Counsels parent about appropriate age to begin testing
      - > 18 years, or
      - 5-10 years before youngest diagnosis
    - Age appropriate counseling for child
Summary of scenario

• Role of GC
  – Cancer risk assessment
  – Create a family testing plan
  – Pre-testing counseling and informed consent
  – Coordinate genetic testing
  – Communicate cancer risks in HBOC
  – Explain inheritance
  – Communicate guidelines and management options for HBOC
  – Psychosocial support and counseling
  – Resources

www.nchpeg.org/hboc/products/case-unsuspecting-male-%E2%80%93-marcus
Resources for You and Your Patients

www.nsgc.org

Experience the value of a genetic counselor. Increase patient satisfaction and enhance the services you provide to your patients.

The role of genetic testing in healthcare grows more prevalent and complex everyday. Do you have the best resources to provide the most comprehensive, personalized care for your patients?
Resources for You and Your Patients

- American Board of Genetic Counseling
  www.abgc.net

- National Coalition for Health Professional Education in Genetics (NCHPEG)
  www.nchpeg.org

- March of Dimes: Genetics and Your Practice
  www.marchofdimes.com/gyponline/index.htm2
Summary

• Genetic professionals have specialized skills in managing genetic risks that can benefit your patients

• Spectrum of roles for PCP in genetic counseling
  – direct referral
  – collaborative care
  – PCP genetic evaluation and counseling
Thank you for your participation!

For more information, please contact

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www.GeneticsinPrimaryCare.org