Genetic Counseling: Case Studies

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Outline

- Genetic Counseling Overview
- Role of a Genetic Counselor in an HTC
- Things to Consider if YOU are in a Genetic Counseling Role
- Case Examples
- Available Resources
What is Genetic Counseling?

A non-directive, educational process helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

- Providing education about:
  - Individual risk
  - Inheritance
  - Testing options
  - Family planning options
  - Available resources
  - Research

- Identifying and address psychosocial issues relating to genetic testing and diagnosis

- Helping individuals and/or families:
  - Understand available options
  - Reach decisions based on their personal values and feelings
  - Adjust to complex information, uncertainties or new diagnoses
  - Facilitate communication with appropriate relatives
Who is a Certified Genetic Counselor?

- Health professional
- Completed a 2-year Master’s degree in genetic counseling
  - ~30 accredited programs nation-wide
  - Training in medical genetics and psychosocial counseling
- Certified through exam by American Board of Genetic Counseling
- Continuing education requirements to remain certified

- Licensed profession (state dependent)
Genetic Counselor Scope of Practice
(per PA state licensure)

- Provide genetic counseling to clients, which includes:
  - Obtain and evaluate individual and family medical histories to determine genetic risk
  - Discuss features, natural history, means of diagnosis, genetics and environmental factors and management of risk for the genetic or medical conditions
  - Identify and coordinate genetic laboratory tests and other diagnostic studies
  - Integrate results of genetic tests and other diagnostic studies with personal and family medical history to assess and communicate risk factors
  - Explain the clinical implications of genetic laboratory tests and other diagnostic studies and their results
  - Evaluate patient/family responses to the condition or occurrence risk and provide client-centered counseling
  - Identify and utilize resources to provide medical, educational, financial and psychosocial support/advocacy
  - Provide written documentation of medical, genetic and counseling information for families and health care professionals

- Indications requiring diagnosis/treatment outside the scope of practice should be referred to an appropriate health care practitioner
The Genetic Counselor at the HTC

- Document family history, with special focus on bleeding/clotting symptoms
- Help individuals and families understand the inheritance of their bleeding disorder
- Provide education about the individual or family bleeding disorder diagnosis
- Identify at-risk family members and discuss appropriate testing options available for them

- Support and facilitate family communication and evaluation of relatives
- Explain genetic testing that is available, including clinical utility, benefits, and limitations
- Identify and coordinate the best genetic test available and the ideal testing strategy within the family
- Review genetic testing results with the individual/family and provider

- Discuss psychosocial issues and responses to hereditary or genetic information
- Review reproductive and family planning options
- Participate in research efforts relating to genetics
- Support the multi-disciplinary team
Things to Consider if YOU are in a Genetic Counseling Role

- Psychological Responses to Genetic Information
  - Lived Experience of Being a Carrier and Mother of a Child with Hemophilia

- Informed Decision Making in Genetic Counseling

- Facilitating Family Communication
  - Communication within Hemophilia A Families
Psychological Responses to Genetic Information

- Information may psychologically overwhelm patient
- Common emotional responses:
  - Denial
  - Anger
  - Guilt
  - Shame
  - Grief
  - Fear
  - Apprehension
- GC role is to provide supportive environment to express and process through these responses
The Lived Experience of Being a Carrier and Mother of a Child With Hemophilia

<table>
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<tr>
<th>Time After Diagnosis</th>
<th>Turning Point</th>
<th>Reconciliation with Changing Life</th>
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<td>• Feeling sad about the child’s illness</td>
<td>• Capturing crucial knowledge</td>
<td>• Reclaiming a sense of security</td>
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<td>• Sense of being accused</td>
<td>• Sharing the burden and sorrow with others</td>
<td>• Adapting professional life to the new situation</td>
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<td>• Unable to protect the child from suffering</td>
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<td>• Changing and growing as a person from the experience</td>
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<td>• Overwhelmed by worries and fear for the child</td>
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<td>• Being hopeful</td>
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<td>Most women did not fully realize their status/risk until their child was</td>
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Informed Decision Making in Genetic Counseling

- Emphasis on patient autonomy
- Education
  - Features, natural history and range of variability of condition
  - Genetic basis
  - Diagnosis and management options
  - Chance of occurrence/recurrence
  - Possible economic, social, and psychological impacts
  - Resources available
- Complete disclosure of information
- Non-directive counseling
- Recognize influences
  - Knowledge, Misconceptions, Religion, Finances, Family dynamics, Support systems
- Anticipatory guidance
- Supportive environment
Family Communication

• Knowledge is power!

• Communication may lead to earlier diagnosis and prevention of complications in relatives

• Communication within Hemophilia A Families
    ▪ Information is selectively communicated
    ▪ Concerns about whether to tell, what to tell, who to tell, when to tell
  ○ Gregory et al (2007)
    ▪ Differences in communication to obligate and non-obligate carriers
    ▪ Sisters may have better understanding of possible carrier status than daughters
    ▪ Readiness to receive information is variable and depends on various factors
Family Communication (cont.)

- Tools available
  - ‘Genetic Counseling for Inherited Bleeding Disorders’ brochure
  - Family letter template
  - Fact Sheets
  - At-risk relative identified card

To Whom It May Concern:

Your relative, John Doe, recently had genetic testing for hemophilia A/B. John tested positive for gene change in the factor VIII gene, which causes hemophilia A. If John has a female relative, you may be a carrier of hemophilia and have a son with the condition. Genetic testing can tell you for certain if you are a carrier or not. This letter will summarize your genetic test results and what they mean for you.

**TEST RESULTS:** The lab found a change in John’s factor VIII gene, which causes hemophilia A. This technical name for the gene change is a “heterozygote” at the DNA level and is “hemophilia A” at the protein level. The testing was performed at the Laboratory North in City, State.

**HOW HEMOPHILIA RUNS IN THE FAMILY:** If a female has a factor VIII gene change, she is a carrier of hemophilia. If she has only one copy of hemophilia in her DNA, but they can have a son with hemophilia. There are four possibilities with each pregnancy to transmit the carrier of hemophilia:
1. A 25% chance (1 in 4) of having a male with hemophilia
2. A 25% chance of having a male who does not have hemophilia
3. A 50% chance of having a female who is a carrier of hemophilia
4. A 25% chance of having a female who is not a carrier

You may be interested in genetic testing in order to find out if you are a carrier of hemophilia. This specialized care is important in hemophilia and they are well enough to make the genetic counseling easier to understand and discuss. However, it is important to keep in mind testing in a younger relative who will be undergoing elective surgery or if you are planning to undergo medical procedures that may cause bleeding is always medically necessary, because her doctor may change her medical care.

**GENETIC TESTING:** Since we inherited factor VIII gene changes that cause hemophilia in John, doing genetic testing is very important and may require a small blood sample. We recommend you consult with a genetic specialist before doing any testing, so you know what carrier testing may be helpful and what it means if you test positive. Please share this letter with your primary care provider and he/she can refer you to a genetic specialist.

*If you have any questions about the letter or genetic testing for hemophilia please do not hesitate to call me. My direct phone number is __________. My email address is __________.*

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**Hemophilia Carrier Fact Sheet**

**WHAT DOES IT MEAN TO BE A “CARRIER” OF HEMOPHILIA?**
- Usually, males have 1 normal copy of both the factor VIII gene and the factor IX gene. A male with hemophilia A or B has no abnormal copy of one of these genes.
- Usually, females have 1 normal copy of both the factor VIII gene and the factor IX gene. **A “carrier” of hemophilia A or B is a female who has 1 abnormal copy of one of these genes.**

**WHY IS IT IMPORTANT TO FIND OUT IF YOU ARE A CARRIER?**
- It is important medical information for you.
  - Carriers can have complications, leading to health issues and type of bleeding.
  - Carriers may need a special medical management plan for high risk surgery, such as surgery or childbirth.
- It is important medical information for your family.
  - Carriers have 25% chance (1 in 4) of having a child with hemophilia. This means that the abnormal copy will be passed on to the next generation. A “carrier” of hemophilia A or B is a female who has 1 normal copy of one of these genes.

**HOW CAN YOU FIND OUT IF YOU ARE A CARRIER?**
- Get a blood test.
  - Your blood is analyzed to see if it contains any abnormal copies of the factor VIII gene.
  - You have 1 out of 3 chances of being a carrier.

**Importantly, this is a blood test that is not affected by whether you are a carrier or not.**

**WHAT DOES IT MEAN TO BE A “CARRIAGE” OF HEMOPHILIA?**

- You may have questions about this information or want to find out if you are a carrier, please contact the Hemophilia Center of Western Pennsylvania at 1-205-725-7252.
Case Examples

- Available in print at conclusion of presentation
Resources

- My Life, Our Future Genetic Counseling Guidelines
- NHF MASAC Guidelines
- National Society of Genetic Counselors [www.nsgc.org](http://www.nsgc.org)
- Local NHF Chapter
- Genetic Information Non-Discrimination Act
Questions