GENETIC COUNSELING RESOURCES

Mid-Atlantic Region III Annual Meeting
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Outline

• Genetic Counseling Overview
• Organizations Addressing Genetics in Bleeding Disorders
• Genetics Tools for Providers in Bleeding Disorders
• General Genetics Resources
Genetic Counseling Overview

- **Genetic Counseling**
  - The process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease
  - Includes:
    - 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

- **Genetic Counselors**
  - Health professionals
  - Master’s degree in genetic counseling
  - Training in medical genetics and psychosocial counseling
## Organizations Addressing Genetics in the Bleeding Disorder Community

<table>
<thead>
<tr>
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<th>NHPCC Genetics Working Group</th>
<th>MAR III Genetics Sub-Committee</th>
<th>NHF Genotyping Working Group</th>
<th>NSGC</th>
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</thead>
<tbody>
<tr>
<td><strong>Members</strong></td>
<td>ATHN representative, regional coordinator, GCs, consumers</td>
<td>Regional coordinator, physician, SW, GC, consumer</td>
<td>Consumers, NHF chapters, physicians, GCs</td>
<td>Genetics professionals</td>
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<tr>
<td><strong>Purpose</strong></td>
<td>Develop and implement a process for collaborating with and providing genetics related materials to the regional networks, consumers, and stakeholders</td>
<td>Facilitate gathering, planning and dissemination of information about genetic services using the Life Course perspective to HTC staff and consumer groups in the region</td>
<td>Provide NHF with guidance in the development and outreach of the My Life, Our Future project</td>
<td>Advancing the roles of genetic counselors in health care by fostering education, research, and public policy to ensure availability of quality genetic services</td>
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<td><strong>Completed Projects</strong></td>
<td>• Genetic Counseling for Inherited Bleeding Disorders Brochure</td>
<td>• Genetics Information Toolkit</td>
<td>• MLOF Genetic Counseling Checklist</td>
<td>• Position Statement on Family Health History in the Electronic Health Record</td>
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<td><strong>Intended Areas of Future Work</strong></td>
<td>• Pre-/post-test FAQs for genotyping • Family planning brochure • Outreach to NSGC</td>
<td>• Evaluation and annual update of the Genetics Information Toolkit • Support of NHPCC demonstration project • Support of FRUIT tool</td>
<td>• Educational outreach to the HTCs and patients who may not have readily available genetic counseling services</td>
<td>• “Update on Bleeding Disorders” session at 2014 NSGC Annual Meeting</td>
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Genetics Tools for Providers in Bleeding Disorders

- CDC Hemophilia A Mutation Project (CHAMP) and CHBMP
  - Excel database containing single listing for each \textit{F8} (CHAMP) and \textit{F9} (CHBMP) mutations reported worldwide from publicly available databases and publications
  - Each mutation is uniquely identified using HGVS nomenclature for DNA and predicted protein changes
  - Includes severity, factor level, inhibitor history, and references to first reports
  - \url{http://www.cdc.gov/ncbddd/hemophilia/champs.html}

- Genetic Counseling Brochure (NHPCC)
  - Information about who may benefit from genetic counseling and how a genetic counselor may be helpful

- Find a Genetic Counselor (NSGC)
  - Directory developed to assist health care providers and patients in locating genetic counseling services
  - Can be searched by State, City, Name, Institution, Work Setting, Type of Specialty or Zip Code
  - \url{http://nsgc.org/p/cm/ld/fid=164}

- My Life, Our Future Genetic Counseling Guidelines (NHF WG)
  - \url{www.athn.org}, MLOF resources

- Tools for Facilitating Family Communication (UVA, HCWP)
  - Family Letter Template
  - At-Risk Relative Identification Card
My Life, Our Future
Genetic Counseling Guidelines

These guidelines are intended to support adequate pre- and post-test genetic counseling of patients with a personal or family history of hemophilia. Each check box topic should be addressed in some degree; however, the level of detail appropriate for each topic may be determined on a case-by-case basis.

POST-TEST

☐ Disclose results
  • Should be done by the same individual who did pre-test counseling
  • Stick with the original plan, including predetermined time and location/method
  • Review the result
  • Explain what it means for the individual’s personal risk
    ○ Confirms hemophilia diagnosis vs. carrier vs. not carrier vs. uncertain
  • Explain what it means for other family members’ risks
    ○ Who else is at-risk or not at-risk based on these test results (i.e. siblings, children?)

☐ Assess and discuss psychosocial impact of results
  • Surprising or not surprising?
  • Concerning or not concerning?
  • Other feelings experienced?

☐ Review utility of test result (dependent on individual and his/her level of interest)
  • For all individuals:
    ○ Share information with relatives
    ○ Consider reproductive planning options, including:
      ▪ No changes to reproductive planning — accept associated risks and plan accordingly for delivery
      ▪ Prenatal testing — most reliable if familial mutation is known, must be done in approved center
        - Chorionic Villus Sampling
        - Amniocentesis
      ▪ Pre-Implantation Genetic Diagnosis (PGD)
      ▪ Sperm, egg or embryo donation
Tools for Facilitating Family Communication

To Whom it May Concern:

Your relative, John Doe, recently had genetic testing for hemophilia A/B. John tested positive for a gene change in the factor VIII gene, which causes hemophilia A/B. As John’s 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 John’s 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/B in males. The technical name of the gene change is “c.1215delC” in the DNA level and “p. N749fs” at the protein level. The testing was performed by the Laboratory Name in City, State.

HOW HEMOPHILIA RUNS IN THE FAMILY: If a female has a factor VIII gene change, she is a carrier of hemophilia. Most females who are carriers of hemophilia are healthy, but they can have a son with hemophilia. There are four possibilities with each pregnancy in females who carry 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 25% 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. We typically do not recommend carrier testing in females until they are old enough to make the decision themselves (usually as teenagers and older). However, it may be important for genetic testing in a younger female who will be undergoing elective surgery or if she is showing signs of easy bleeding or heavy menstrual bleeding, because her doctor may change her medical care.

GENETIC TESTING: Since we know the factor VIII gene change that causes hemophilia in John, doing genetic testing in you is very simple and only requires a small blood sample. We recommend for you to meet with a genetics specialist before doing any testing, so you know why 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 genetics specialist.

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

Sincerely,

Name
Title

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Hemophilia A is a genetic disorder. It is caused by a gene change, which can be passed from generation to generation. Your diagnosis means that other males and females in your family may carry a gene change that can cause them to have hemophilia A. We encourage you to share this information with your family members, because it can provide them with valuable information for their own healthcare.

Specifically, members of your family who may currently benefit from risk assessment include:

{Name, Age, Relation}

Education and evaluation of these individuals may be significantly beneficial for themselves, as well as their offspring. We encourage your relatives to contact us at 412-209-7280 with any questions or to set up an evaluation. If your relatives do not live in this area, we can likely provide them with contact information for a provider who can offer them these services locally.

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Family Letter Template (UVA)

At-Risk Relative Identification Card (HCWP)
Patient Education Materials

Genetic Counseling for the Inherited Bleeding Disorders Community

Genetic Counseling Brochure (NHPCC)

Carrier Fact Sheet (HCWP)

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 an abnormal copy of one of those genes.
- Usually, females have 2 normal copies 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 those genes.

WHY IS IT IMPORTANT TO FIND OUT IF YOU ARE A CARRIER?
- It is important medical information for you:
  - Carriers can have symptoms, including heavy periods, easy bruising and prolonged bleeding
  - Carriers may need a special medical management plan for high-risk events, such as surgery or childbirth.
- It is important medical information for your family:
  - Carriers have a 50% chance to pass the abnormal copy to their children. Males who inherit the abnormal copy will have hemophilia. Females who inherit the abnormal copy will be carriers.
  - If you are not a carrier, you cannot pass an abnormal copy to your children.
  - Male babies who are at risk for hemophilia:
    - May benefit from a special delivery plan.
    - Can be tested at birth using a cord blood kit. (These are available at our center.)
    - May benefit from delaying surgery, including circumcision, until test results are known.
  - Determining your carrier status could help identify others in your family who may be at risk.
- It provides information to use in the future:
  - It may help you make family planning decisions.
  - It may enable prenatal diagnosis, if desired.

HOW CAN YOU FIND OUT IF YOU ARE A CARRIER?
- You are a carrier if:
  - Your father has hemophilia.
  - You have 1 son who has hemophilia and at least 1 other male relative who has hemophilia.
  - You have 2 or more sons who have hemophilia.
- You may be a carrier if:
  - You have 1 son who has hemophilia and no other male relatives who have hemophilia.
  - You are the sister, aunt, grandmother, niece or cousin of someone who has hemophilia.

Two kinds of testing can be performed:
- Genetic testing. This is a blood test that looks at either your factor VIII gene(s) or factor IX gene(s) to see if they are normal or abnormal. This is the best test to find out if you are a carrier or not. In a family, genetic testing is most useful if a male with hemophilia has this testing first. If your carrier status can be determined based on your family history, you likely do not need this test.
- Factor level. This is a blood test that measures factor VIII or factor IX levels in the blood. This test is less accurate than genetic testing for determining a person’s carrier status. A factor level is most useful to help predict if a known carrier may develop symptoms. This test cannot confirm carrier status. If this test is the only one you have had done, additional testing may be necessary to provide accurate information about your carrier status.

If you have questions about this information or want to figure out if you are a carrier, please contact the Hemophilia Center of Western Pennsylvania at 412-209-7411.
General Genetics Resources

  - Expert-authored, peer-reviewed disease descriptions presented in a standardized format and focused on clinically relevant and medically actionable information on the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions

  - Consumer-friendly information about the effects of genetic variations on human health

• **Genetic Alliance**  [http://www.geneticalliance.org/about/webinararchive](http://www.geneticalliance.org/about/webinararchive)
  - Nonprofit health advocacy organization committed to transforming health through genetics and promoting an environment of openness centered on the health of individuals, families, and communities
  - Open space for shared resources, creative tools, and innovative programs

• **National Society of Genetic Counselors**  [www.NSGC.org](http://www.NSGC.org)
  - Professional organization with the mission of advancing the various roles of genetic counselors in health care by fostering education, research, and public policy to ensure the availability of quality genetic services
  - Discussion of all issues relevant to human genetics and the genetic counseling profession