Role of Genetic Counseling in FH: What Referring Physicians Need to Know

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Disclosures

- Nothing to disclose.
Introduction to the topic

- Who are genetic counselors?
- What is the role of the genetic counselor in patients with FH?
- What is involved with genetic counseling?
- Case example
- Family issues
- Genetic counseling resources
Genetic Counselors
Genetic Counselors

- Master’s-trained healthcare professionals with expertise in genetics and psychosocial counseling
- Training guided by practice-based competencies developed by the American Board of Genetic Counseling (ABGC)
  - Curriculum - Basic science, medical genetics, counseling
  - Clinical internships - Variety of settings including cancer, reproductive, pediatric, cardiovascular, labs, others
- Certification examination administered by the Accreditation Commission for Genetic Counseling (ACGC)
- 14 states currently issuing licenses with 4 states with bills passed/in rulemaking
What are possible roles for genetic counselors in FH?

- Pedigree collection
- Risk assessment
- Genetic counseling
- Genetic counseling ≠ genetic testing
- Cascade screening implementation
- Family evaluation and coordination
- Discussing screening, prevention, and medical management options
- Education
  - Medical and lay audiences
- Outreach and Awareness
- Part of multidisciplinary team for comprehensive care
Taking an Informative Pedigree

- ≥3 generations
- 1st degree relatives (children, siblings, parents) and 2nd degree relatives (nieces/nephews, aunts/uncles, grandparents)

Questions should include:
1. Current ages
2. Health status and age at diagnosis
3. Age and cause of death
4. Family’s origin or racial/ethnic background
Example FH Pedigree
Genetic Counseling Appointment

- Medical history
- Family history
  - Collection and review of family members’ medical records
- Physical examination – medical geneticist
- Risk assessment
- Education
- Genetic testing
  - Informed consent, blood draw, insurance preauthorization
- Genetic test result interpretation and disclosure
- Screening/management recommendations in collaboration with referring physician
Genetic Counseling: Additional Services

- Counseling for psychosocial issues related to hereditary disease, genetic testing and results, etc.
- Referral to support groups and advocacy organizations
- Connection with families with the same condition
- Option of DNA storage for future use of patients, families and possibly researchers
- Option to participate in genetics-related research studies
- Workup and evaluation of at-risk family members

Cascade screening
Cascade Screening: Critical for FH Families

- Process of systematic family tracing to identify at-risk relatives
- Testing lipid levels in all FDRs of FH patients
- If the disease-causing mutation has been identified, genetic testing may also be a part of cascade screening
- Newly identified FH cases provide additional relatives who should be screened
- Facilitates early detection and treatment

After FH has been diagnosed, any family member can have lipid or genetic testing, but a cascade strategy saves resources and headaches!

www.fhjourneys.com
Letter to referring physician

- Family History Summary and Assessment
- Genetic Risk Assessment
- Genetic Testing Results (if applicable)
- FH Key Points
- Resources
- Recommendations and Plan
  - Specifics on cascade screening
- Enclosures
  - Pedigree
  - Dear Family Member FH Letter
Dear Family Member,

I have been diagnosed with familial hypercholesterolemia (FH). FH is a genetic disorder characterized by high cholesterol levels in the blood and early heart disease. Based on my diagnosis, you are at risk for also having FH. You could have FH without any physical signs or symptoms and may feel and look healthy. If diagnosed, FH can be treated through lifestyle changes (not smoking, regular exercise and a healthy diet) and medication. You can determine if you have FH by seeing your doctor and having a fasting lipid profile. This is a panel of blood tests that provides information about cholesterol levels. Individuals with FH have high levels of low-density lipoprotein (LDL) cholesterol. Genetic testing for FH is available, however, is generally not needed for diagnosis or management but may be useful in some cases.

FH is passed down through families in an autosomal dominant manner and can be inherited from a parent. All first-degree relatives (parents, siblings, children) of an affected person with FH have a 50% chance of also having FH. If you do have FH, each of your children has a 50% chance of having it as well. If you do not have FH, you would be at the general population risk for developing high cholesterol, and your children are not at risk to have inherited FH.

Genetic Counseling

Genetic counselors are healthcare professionals who can help you understand the inheritance of FH and what this diagnosis means for you and your family. Genetic counselors can also facilitate genetic testing and interpret results. To locate a genetic counselor in your area, ask your doctor or use the search tool available on the website for the National Society of Genetic Counselors, [www.nsgc.org](http://www.nsgc.org).

Learn More About FH

- The FH Foundation ([http://www.thefhfoundation.org/](http://www.thefhfoundation.org/))
- Preventive Cardiovascular Nurses Association ([http://pcna.net/patients/familial-hypercholesterolemia](http://pcna.net/patients/familial-hypercholesterolemia))

Next Steps

Ask your family doctor or primary care physician to check your cholesterol

- A diagnosis of FH is probable if:
  - LDL is greater than 170 mg/dL in adults 20 years of age and older
  - LDL is greater than 155 mg/dL in children and adolescents less than 20 years of age (Williams RR et al. *Am J Cardiol.* 1993;72(2):171-176)

Take a copy of this letter with you when you meet with your doctor. If you are diagnosed with FH, share this letter with your at-risk parents, siblings and children.
Genetic counseling promotes family communication
Powerful information

“Your timing was good. My brother would have ignored his chest pain last night without my earlier call. He's not had an MI but he's in the ED waiting for his stress test. His BP was 188/112 when the squad came. THANKS!”
Costs

- Genetics consultation appointment
  - Typically covered by insurance
  - Specialist co-pay
- **LDLR** sequence analysis
  - ~$1300
- **LDLR** deletion/duplication
  - $560
- FH “Comprehensive Evaluation”
  - (**LDLR** sequence analysis and **APOB** partial gene sequence analysis with reflex option to **LDLR** deletion/duplication and reflex option to **PCSK9**)
  - ~$3,500
- Family specific single site testing
  - $200-400
Case example
Patients KS and CK

- Father-daughter pair who attended Cardiovascular Genetic Medicine Program clinic visit together
- Longstanding diagnoses of FH
- KS participated in MEDPED program registry (Make Early Diagnosis to Prevent Early Deaths)
  - Research project and nonprofit organization funded to register and help treat people with inherited cholesterol disorders
  - Dr. Roger Williams, founder of the Inherited High Cholesterol Foundation, died in the airplane crash of Swissair Flight 111 on September 2, 1998
The diagnosis of FH in this family is not ambiguous, so genetic testing was not pursued.

Points discussed in detail:
- Autosomal dominant inheritance (did not know)
- Cascade screening recommendations – including pediatric patients
- FH is a treatable condition
  - If family members have FH they require immediate initiation of treatment and aggressive management of other CHD risk factors (e.g. hypertension, diabetes, smoking)
  - Urged family members not currently undergoing treatment to seek medical therapy immediately
  - Urged family members who are smokers to quit
- Patients who do not respond adequately to, or are intolerant of, initial statin therapy should be referred to a lipid specialist
- For children with FH, either consultation with or referral to a lipid specialist for management is recommended

Provided family letter

Reviewed online resources and upcoming FH registry
Managing Duty to Warn vs. Privacy

- The concept of genetic malpractice was first discussed in the 1970s revolving around the lack of consideration to family members at risk of an inherited disease.

- *JAMA.* The “duty to warn” a patient’s family members about hereditary disease risks. 2004

- **We need** to consider not only the patient in front of us, but also the other family members at risk for FH.

- Patient must be informed that other family members are at risk.

- Encourage sharing information among family members.

- Provide letters/literature for sharing with family.

- Document conversation about sharing information and recommendations for screening family members.
How are we able to share genetic information?

B. **Family Members**
My family members may wish to have access to records from my risk assessment appointment including my appointment letter, my family tree, and my genetic testing results. I authorize release of these records to the individuals listed below should they request it. They may also request this information if I am deceased or otherwise unable to consent for release of my records and/or test results.

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All of my blood relatives and/or their healthcare providers

**YES** (initials/date) **NO** (initials/date)

Signature of patient authorizing release

Date
Family Issues

- Encourage family involvement and a shared approach to decision making
- Common emotional issues
  - Grief, Guilt, Blame, Responsibility
- Survivor guilt
  - Need to prepare family members for discordant results
  - How do you “break the news” when you are testing multiple people in a family simultaneously?
- Take care to avoid coercion by relatives
- Protect privacy
- Paternity
Genetic Discrimination

- Genetic Information Non-discrimination Act (GINA)
  - Federal Law signed on May 21, 2008
  - Prevents health insurers from denying coverage, adjusting premiums, or otherwise discriminating on the basis of genetic information
    - Group and self-insured policies
  - Employers cannot use genetic information to make hiring, firing, compensation, or promotion decisions
  - No protection for disability or life insurance discrimination
To summarize…
What evidence recommends and supports genetic counseling in cardiovascular genetics?

- A number of expert opinions recommend genetic counseling for individuals and families with hereditary heart conditions. (HRS/EHRA 2011 expert consensus statement, Judge et al 2009, Fowler et al 2009)

- The Heart Failure Society of America recommends genetic counseling for patients and families with cardiomyopathy. (Hershberger et al 2009)

- Genetic counseling interventions have been shown to increase the number of at-risk relatives with hereditary heart disease who present for recommended clinical evaluations. (Forrest et al 2008, Van Der Roest et al 2009)

- Research has also demonstrated that families with inherited cardiac conditions prefer having genetic counseling as part of their care and have better psychological outcomes when a genetic counselor is involved in their care. (Ingles et al 2008, Andersen et al 2008)

- Genetic counseling can also be key to ensuring the accurate and appropriate use and interpretation of genetic tests. (Caleshu et al 2010, Brierly et al 2010)
“We strongly advocate the involvement of physicians and centers with expertise in cardiovascular genetics…They will provide genetic counseling.”
For Healthcare Providers

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?

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 about genetic counseling
- Find a genetic counselor near you
- Understand your family history
- Tools and resources for your genetic health

For Genetic Counselors

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

- Explore a career as a genetic counselor.
- Learn about the skills of genetic counselors.
- Experience the benefits of NSGC membership.
- Learn more about NSGC’s resources for genetic counselors.
- Hear from the NSGC President!

nsgc.org

Active Online Courses

If you have purchased any of the NSGC online educational courses your access link(s) will be listed below. Please Note: there may be a 5-10 minute delay before you can access your course after purchase.
Summary and Take Home Messages

- Genetic counselors are specialized health care professionals with expertise in genetics and counseling.
- A pedigree should be collected, assessed, and stored on every patient with FH.
- Cascade screening needs to be performed.
- There may be family psychological issues when dealing with genetic conditions.
- Genetic counselors are available to you as a resource.