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

Amy C. Sturm, MS, LGC
Associate Professor
Division of Human Genetics
Department of Internal Medicine

May 2, 2014
Disclosures

- Nothing to disclose.
Topics covered today

- What is genetic counseling?
- Who are genetic counselors?
- What are the roles of genetic counselors for patients with FH?
- What is included during genetic counseling?
- Family dynamics
- Genetic discrimination
- Case example
- Genetic counseling resources
What is genetic counseling?

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

- Genetic counseling has been recognized by physicians for its many positive contributions for patients and families with inherited forms of heart disease and is indicated regardless of whether or not genetic testing will be performed.

- Multiple genetic counseling service delivery models exist, including in-person, telephone, group, and telegenetic, as well as varied types of referral patterns of patients for genetic counseling.

Genetic Counselors
Who are 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
- 19 states currently issuing licenses with 5 states with bills passed/in rulemaking
Figure 4. Primary Work Setting

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

*“Other work settings” include: Not-For-Profit Organization, University/Non-Medical Center, Diagnostic Laboratory – Academic, Government Organization or Agency, Federal/State/County Office, Research Development/Biotechnology Company, Pharmaceutical Company, Health Advocacy Organization, Internet/Website Company, Private Practice - Self-Employed, Professional Organization, Bioinformatics Company, Outreach/Satellite/Field Clinic.*
What are possible roles for genetic counselors in FH?

- Pedigree collection and risk assessment
- Genetic testing
  - Pre- and post-test genetic counseling
- Cascade screening implementation
- Family evaluation and coordination
- Discussing screening, prevention, and medical management options in conjunction with managing physician
- Provision of resources from national organizations and advocacy groups
- Coordinating research protocols
  - Enrolling FH patients into CASCADE FH Registry
- Education
  - Medical and lay audiences
- Outreach and Awareness
- Part of multidisciplinary team for comprehensive care
Constructing an Informative Pedigree

• >3 generations

Questions should include:
1. Current ages
2. Health status and age at diagnosis
3. Age and cause of death
4. Focus on “red flags” (e.g. premature CAD, SCD)
5. Family’s racial/ethnic background

Family history is imperative in:
1. aiding diagnosis
2. determining inheritance pattern
3. identifying at-risk relatives
4. selecting the most informative family member for genetic testing initiation
The Process of Genetic Counseling

• Medical history
• Family history
  • Collection and review of family members’ medical records
  • Cholesterol panels, other medical records, death certificates, autopsy reports
• Physical examination – collaborating physician
  • Evaluation for physical signs of FH
• Risk assessment
  • Inheritance, recurrence risk
• Education
• Genetic testing
  • Informed consent, sample collection, insurance pre-authorization
• Genetic test result interpretation, disclosure, and documentation
• Screening/prevention/management recommendations in collaboration with referring HCP
• Discussion of reproductive options
Genetic Counseling: Additional Services

- Psychosocial counseling and anticipatory guidance
- Provision of resources from national organizations and advocacy groups
- Connection with families with the same condition
- Discussion of DNA banking availability, when applicable
- Discussion of available research options
  - Enrolling FH patients into the CASCADE FH Registry
- Facilitation of family-based care/evaluation of at-risk family members

Cascade screening
Cascade Screening: Critical for FH Families

- Process of systematic family tracing to identify at-risk relatives
- Should begin with 1st-degree relatives and then extend to 2nd and 3rd degree in a stepwise fashion
- DNA testing should be used if the pathogenic mutation has been identified
- Newly identified FH cases provide additional relatives who should be screened
- Facilitates early detection and treatment
- A cost-effective method for identifying people with FH

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

Nordestgaard et al. European Heart Journal. 2013
CDC Office of Public Health Genomics has classified cascade testing for FH as a “Tier 1” genomic application.

FH is a “winnable battle”
Genetic counseling summary letter to referring physician and patient

- Medical and 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
  - CASCADE FH Registry brochure
  - Copy of genetic testing results (if applicable)
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.

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.* 1999;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
Costs

- Genetics consultation appointment
  - Often covered by insurance
  - Specialist co-pay
- **LDLR** sequence analysis
  - ~$1300
- **LDLR** deletion/duplication
  - ~$560
- **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
Genetic counseling promotes family communication
Family Dynamics

- Encourage family involvement and a shared approach to decision making
- Common emotional issues
  - Grief, Guilt, Blame, Responsibility
- Parental guilt
  - May be helpful to emphasize benefits of information (i.e. Knowledge is Power)
- 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
  - “Nagging” of children by parents; could lead to relationship breakdown
- Protect and maintain privacy and confidentiality
- Paternity
Managing Duty to Warn vs. Privacy

- The concept of “genetic malpractice” revolves 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

- FH index cases should be provided with written information and be encouraged to give this to their relatives*
  - Clearly explains testing process and implications
  - Written generally to avoid alarm and concern
  - Emphasize health consequences of missed FH diagnosis and health gains of diagnosis and treatment

- Document conversation about recommendations to share information with family and for family member screening

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
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
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

Review online resources
Refer to CASCADE FH Registry
Provide Dear Family Member Letter
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)
AHA Policy Statement

Genetics and Cardiovascular Disease
A Policy Statement From the American Heart Association

Euan A. Ashley, MRCP, DPhil, Chair; Ray E. Hershberger, MD, Co-Chair; Colleen Caleshu, ScM, CGC; Patrick T. Ellinor, MD, PhD, FAHA; Joe G.N. Garcia, MD; David M. Herrington, MD, FAHA; Carolyn Y. Ho, MD; Julie A. Johnson, PharmD; Steven J. Kittner, MD, FAHA; Calum A. MacRae, MD, PhD; Gia Mudd-Martin, RN, MPH, PhD; Daniel J. Rader, MD, FAHA; Dan M. Roden, MD, FAHA; Derek Scholes, PhD; Frank W. Sellke, MD, FAHA; Jeffrey A. Towbin, MD, FAHA; Jennifer Van Eyk, PhD; Bradford B. Worrall, MD, MSc, FAHA; on behalf of the American Heart Association Advocacy Coordinating Committee

“We strongly advocate the involvement of physicians and centers with expertise in cardiovascular genetics…They will provide genetic counseling.”
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?
NSGC CV SIG Directory 2014 – Google Maps
Who to Refer for Genetic Counseling

- Patients with a known diagnosis of FH
- Patients with a possible diagnosis of FH
- Patients with premature CAD and hypercholesterolemia
- At-risk relatives of FH probands
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 testing needs to be performed and index patients should be provided with tools to assist them in informing their at-risk relatives about FH.
- Families with FH may face different psychological issues.
- Genetic counselors work as members of the healthcare team and are available to you as a resource.
Genetic counselors can help win the “FH Battle”!

“Our biology does not stop: the risk of developing CHD as a consequence of FH will still be present, even if relatives live in ignorance.”

*Newson and Humphries. EJHG. 13, 401-408 (2005).