GENETICS: INTRODUCTION TO GENETIC COUNSELING

Goal:

Recognize the need for and components of the genetic counseling process

After completing this activity participants will be able to:

- Identify appropriate candidates for genetic counseling
- Describe the basic process and goals of genetic counseling
- Recognize genetic counseling interactions warranting a nondirective approach
- Locate genetic counseling and informational resources

Professional Practice Gaps

In an effort to define what healthcare providers need to know about medical genetics, several organizations developed core competencies (NCHPEG, 2000; ASHG, 2001). However, because clinical genetics is a relatively young and evolving field of medicine, many practitioners received insufficient formal genetics education. As a result, they express a lack of confidence in their clinical genetics knowledge and a lack of confidence in their ability to provide genetic counseling.

GENETIC COUNSELING DEFINED

Genetic counseling is a comprehensive term that describes a communications process from beginning to end. The term was coined by Sheldon Reed in 1947 (Resta 2006). Genetic counseling has since been defined by the American Society of Human Genetics (ASHG). ASHG defines genetic counseling as

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (National Society of Genetic Counselors' Definition Task Force 2006).

Genetic counseling does not include physical exams or diagnosis of genetic conditions. It is strictly a communication process. In situations where a diagnosis has not yet been made, accurate genetic counseling cannot be provided to the patient. Referral to a clinical geneticist and/or other appropriate specialist(s) may be needed. In some cases, a specific diagnosis cannot be made or is not possible to confirm. In this situation, a patient is provided information about possible differential diagnoses and any potential risk estimates.

Genetic counseling may be completed in a single visit, as is often the case for routine prenatal diagnosis counseling sessions for women who will be 35 years or older at delivery. Alternatively, the

communication process may span an individual's lifetime, as is often the case for people diagnosed with genetic conditions.

GOALS OF THE GENETIC COUNSELING PROCESS

In addition to defining genetic counseling, ASHG outlines goals for the process (The National Society of Genetic Counselors' Task Force Report 2006; National Cancer Institute 2014).

ASHG Goals for Genetic Counseling Help the Individual or Family to Do the Following:

- Comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management
- Appreciate the way that heredity contributes to the disorder and the risk of recurrence (occurrence) in specified relatives
- Understand the alternatives for dealing with the risk of recurrence (occurrence)
- Choose the course of action that seems appropriate in view of risk, family goals, and ethical and religious standards, and act in accordance with that decision
- Make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence (occurrence) of that disorder

  (The National Society of Genetic Counselors' Task Force Report 2006; National Cancer Institute 2014)

GENETIC COUNSELING PROVIDERS

Anyone in the healthcare profession can provide genetic counseling. Traditionally, genetic counseling has primarily been offered by genetics professionals; however, it is likely that nongenetics professionals will begin to assume a larger role in the provision of genetic counseling.

Genetics Professionals

- Master's degree professionals certified by the American Board of Genetic Counseling (genetic counselors)
- Physicians certified by the American Board of Medical Genetics (clinical geneticists)
- Nurses with specialized genetics training

In many genetics clinics, genetic counselors and/or genetic nurses work with MD clinical geneticists. Although each clinic is unique, often counselors and nurses provide the bulk of information gathering, risk assessment, and counseling. MD clinical geneticists usually review medical records, conduct physical exams, and attempt to determine or confirm a diagnosis. For genetic counseling indications where diagnostic evaluations are not needed, genetic counselors may be the only provider with whom a patient meets. Genetics clinics with MD clinical geneticists are often located in large academic medical centers or outreach programs affiliated with such centers.

It is important to recognize that genetic counseling is not limited to genetics clinics. It is also often provided in specialty clinics, such as cancer clinics for hereditary cancer syndromes, pulmonary clinics for cystic fibrosis, maternal fetal medicine departments for high-risk pregnancies, and many

others. In addition, genetic counseling is performed in the primary care setting. Depending on the primary care provider's comfort level with and knowledge base in genetics, different degrees of genetic counseling are currently being offered to patients in the primary care setting. However, as genetic knowledge and the number of available tests grow, it is likely that primary care physicians will increasingly encounter opportunities in the clinic to offer genetic counseling. Primary care physicians will undoubtedly face the difficult decision of whether or not to personally manage the growing number of patients for whom genetic counseling will become appropriate. At present, there are no global guidelines or recommendations instructing primary care physicians when a genetics referral is appropriate. Each primary care provider will have to decide on a case-by-case basis which patients to provide genetic counseling to personally and which patients should be referred to a genetics specialist. **Ultimately, referral decisions should take into account one's knowledge base, ability to adequately provide the genetic counseling process in its entirety, and the availability of genetics resources.**

CANDIDATES FOR GENETIC COUNSELING

Appropriate candidates for genetic counseling include patients who are diagnosed with or at risk for genetic conditions and individuals at risk to have a child with a hereditary condition or birth defect.

Examples of Individuals and Couples Who May Benefit From Genetic Counseling

The National Society of Genetic Counselors provides the following list of examples of individuals and couples who may benefit from genetic counseling:

- Persons or families with a history of cleft lip or palate, congenital heart defects, spina bifida, short stature, or other physical birth defects
- Persons or families with genetic disorders such as Down syndrome, Huntington disease, cystic fibrosis, muscular dystrophy, PKU, hemophilia, and other inherited disorders
- Persons or families affected with mental retardation, hearing or visual impairments, learning disabilities, or other conditions that could be genetic
- Persons or families with a history of certain cardiac, cancer, psychiatric, or neurogenetic adult disorders
- Persons with a history of multiple miscarriages, stillbirths, or early infant deaths involving multiple congenital anomalies
- Women age 34 and over who are pregnant or are planning pregnancy
- Pregnant women at high risk due to abnormal MSAFP* or ultrasound screening tests
- Pregnant women concerned about the effects of exposure to medication, drugs, chemicals, infectious agents, radiation, or certain work conditions (this subspecialty is called **teratology**)
- Persons in specific ethnic groups or geographic areas with a higher incidence of certain disorders, such as Tay-Sachs disease, sickle cell disease, or thalassemias

*MSAFP = maternal serum alpha-fetoprotein

(NSGC 1995-2005a) .

THE GENETIC COUNSELING PROCESS

For any condition, the genetic counseling process is time consuming. A great amount of time is spent face-to-face with each patient or family. Information is collected and then communicated to the patient. In addition, healthcare providers spend a significant amount of time both preparing for appointments and researching information after an appointment. Time outside of appointments is commonly used to review medical records and diagnoses, locate available testing, and find educational resources for the patient or family.

Components of the ideal genetic counseling process are displayed in the box below. Although the components are often incorporated in descending order just as they are listed, genetic counseling is a not fixed process. Sometimes genetic counseling components are addressed in a different order or are repeated. Each counseling session will be tailored to the patient, and, depending on the indication for genetic counseling, different aspects of the process are emphasized. For example, genetic counseling for a 37-year-old pregnant woman (without a significant family history) will tend to center around her age-related risk for trisomy, the option of prenatal diagnosis, and procedure-related risks and benefits. In contrast, counseling a family with a 1-week-old baby diagnosed at birth with achondroplasia (the most common form of dwarfism) will likely focus on education about the condition and helping the family to adjust.

The Genetic Counseling Process Includes the Following:

- Information gathering
- Initial risk assessment
- Counseling
    - discussion of identified risks and/or potential risks
    - education about healthcare options (including available diagnostic or testing options) and available resources
- Discussion of diagnosis and/or test results
- Follow-up counseling

(Genetic Alliance 2009; Nussbaum, McInnes, & Willard 2007)

For the busy primary care provider, the amount of time required to provide genetic counseling may be a significant barrier. Some providers may be able to carve out the time to spend with patients for the initial information-gathering and risk-assessment portions of the counseling process, while others may be able to allocate time for the entire process.

INFORMATION GATHERING

One of the most important pieces of the genetic counseling process is the gathering of information. The medical history of both the patient and the patient's family is collected and documented on a genetic pedigree. Much information can be provided by patients and their family members. However, it is recommended that medical diagnoses be confirmed by obtaining medical records. To expedite the genetic counseling process, it is helpful to work with patients and families prior to the appointment to obtain relevant medical records, including any previous genetic test results.

Pedigrees help the healthcare provider identify any traits running through the family. Each pedigree created for genetic evaluation should include at least 3 generations of maternal and paternal family members. On the pedigree, it is important to differentiate biological family members from nonbiological family members (family members who have been adopted or who have married into the family). The distinction is important, since nonbiological family members do not impact the patient's risk for inherited conditions.

Information Recorded on a Genetic Pedigree Includes the Following:

- Each person's date of birth or current age
- The date and/or age of death of any deceased family members
- Information about health problems or unusual physical characteristics
- Any previous genetic test results or evaluations
- Current pregnancies, results of any prenatal testing or ultrasound information, and estimated delivery date
- Miscarriages, stillbirths, infant deaths
- Infertility
- Ethnic background
- Consanguineous relationships

(Bennett 2010)

RISK ASSESSMENT

Once the information-gathering portion of the genetic counseling process is completed, genetic risks to the patient and/or family can be assessed. Often a specific pattern of inheritance or a potential diagnosis is visible on the pedigree. For example, an autosomal dominant or autosomal recessive pattern of inheritance may be evident. On other occasions, there may not be a previous occurrence of the condition, so neither the inheritance pattern nor a potential diagnosis is obvious upon pedigree review.

Medical Family History Red Flags

Individuals with **findings that are uncommon** or who have **unusual presentations of common disorders** should raise suspicion of a genetic etiology. Some examples of red flags in the medical family history are shown below.

- Multiple family members with the same or related condition
- Familial "clustering" of cancers that may have the same etiology (colon cancer and endometrial cancer due to an HNPCC mutation)
- Ethnicity known to be associated with increased risk for specific genetic conditions (sickle cell among those of African descent, BRCA mutations among Ashkenazi Jewish individuals)
- Consanguinity
- Birth defects
- Mental retardation or developmental delay
- Other unusual physical findings (dysmorphic features, abnormal pigmentation, significant short stature)

- A recognized, classic single gene (cystic fibrosis, muscular dystrophy) or chromosomal (Down syndrome, translocation) disorder
- Congenital or early-onset deafness or blindness
- Unexplained neuromuscular conditions, particularly with early onset (movement disorders, ataxia, hypotonia, seizures)
- Symptoms suspicious for metabolic disease (failure to thrive, loss of milestones, unusual odors)
- Reproductive abnormalities (recurrent pregnancy loss, abnormal development, infertility)
- Common conditions with an unusual presentation (earlier onset or increased severity, combined with other unusual findings)
- Rare cancers or tumors
- Sudden premature death in an apparently healthy person

(Adapted from Bennett 2010)

COUNSELING

After information gathering and risk assessment, information is communicated to the patient. Ideally, genetic counseling is performed in a caring and objective manner. Healthcare professionals providing genetic counseling seek to do so without allowing personal feelings to bias facts given to the patient (NSGC 2006). The healthcare provider does not suggest one course of action over another; instead, the goal of the counseling process is to fully educate patients so each patient may choose the option(s) best suited to his/her beliefs, lifestyle, and economic situation. This approach is often referred to as ***nondirective*** or ***value neutral***. On occasion and when professionally appropriate, a more directive approach can be taken (Fineman and Walton 2000). For example, if a patient is diagnosed with a condition preventable when specific healthcare measures are taken prior to symptom onset and for which other family members are unknowingly also at risk, the healthcare provider may attempt to persuade a reluctant patient to alert at-risk family members.

Information Commonly Offered to Patients Includes the Following:

- Symptoms and health problems that clinically affected people may exhibit
- Treatment options for clinically affected individuals
- An explanation of the way the condition is inherited
- Discussion of the chance family members have of developing the condition
- Discussion of the chance family members have of passing the condition on to children
- Diagnostic and research-based testing options
- Prenatal testing and reproductive options
- Contact information for the nearest clinic(s) managing patients with the condition
- Contact information for any support groups or educational organizations
- Brochures or literature tailored to patients

(Johnson and Brensinger 2000; Plunkett and Simpson 2002)

COMPLICATED SITUATIONS IN COUNSELING

For a patient or family member in whom a diagnosis has already been established, the counseling is straightforward. However, on occasion, a diagnosis cannot be determined. In this situation, genetic counseling is much more complicated. Information provided to the patient usually includes possible differential diagnoses and the risks associated with each.

A great deal of time is spent during genetic counseling to make sure the information provided to the patient is understandable. Depending upon the patient's education level and familiarity with the relevant condition(s), medical terminology is carefully "translated" into more comprehensible lay terms.

Genetic Testing

Sometimes, but not always, the genetic counseling process includes a discussion about genetic testing. In some circumstances, genetic testing may have already been performed. In other cases, testing is not available or is not desirable to the patient. When genetic testing is available, appropriate, and the patient is interested in pursuing it, informed consent must first be obtained (Holtzman and Watson 1998). Informed consent for genetic testing is important because such testing carries with it enormous potential ramifications, is often expensive, and usually has limitations.

Patients elect to proceed with genetic testing for many different reasons and in many different circumstances. Genetic tests are commonly ordered to determine the following:

- Carrier status for family planning
- The genetic status of an unborn child at risk for a genetic condition
- A possible diagnosis in a child or adult
- Individuals at risk for a genetic condition who have not yet manifested clinical symptoms
- Newborn infants at risk to develop specific genetic conditions whose symptoms are deemed preventable or treatable

Approaches

During a genetic counseling appointment, healthcare providers often use several different approaches to explain the same information by incorporating discussions, diagrams, and illustrations. Any risk figures provided to patients are commonly presented in several different ways. For example, a couple may be told that they have a 25% risk to have a child with sickle cell anemia. The healthcare provider will likely also discuss the risk figure as a 1 in 4 chance. When possible, brochures or other educational materials are given to the patient. After most genetic counseling appointments with a genetics professional, a detailed letter reviewing all of the information discussed is sent both to the patient and to the referring physician.

DISCUSSION OF GENETIC TEST RESULTS

If genetic testing options are pursued by the patient, test results are also discussed during the counseling process. For more routine genetic testing, such as carrier testing or chromosome analysis for a woman with previous miscarriages, results are usually provided over the telephone. When test results for routine testing come back positive (increasing risk), patients are usually also offered an additional appointment to discuss results in person. For less routine testing, results are generally given face-to-face regardless of the result. An example of an occasion when face-to-face result disclosure is appropriate is predictive testing for an adult-onset condition in a currently symptom-free individual.

Although the limitations of testing should be discussed during the process of obtaining informed consent from the patient, it is important to revisit these limitations during result disclosure. Genetic testing is full of caveats; depending on the test, a "positive" result does not necessarily indicate that a patient is at increased risk. In contrast, because most genetic tests do not have a 100% detection rate, a "negative" result does not necessarily mean that a patient does not have an increased risk.

FOLLOW-UP COUNSELING

Inherent in the genetic counseling process is the exchange of a vast amount of information. Different patients require different lengths of time to absorb information provided and explore available options. Often the information is used by patients to make significant life decisions and requires a great deal of thought and contemplation.

For some patients, additional appointments are helpful.

Follow-Up Appointments May Be Used To

- Review information previously discussed
- Help patients process emotions
- Help patients make decisions
- Provide additional information
- Answer new questions

For patients who experience significant emotional distress, referral to a local mental health professional for long-term counseling is recommended (NSGC 1995-2005b). If available, patients may also benefit from attending a local support group with other individuals or families in similar situations.

INTRODUCTION TO HUNTINGTON DISEASE PRESYMPTOMATIC TESTING

This chapter will follow a patient, Jason Bennett, through a genetic counseling appointment, where he learns about Huntington disease.

Clinical Example

Huntington disease (HD) is an adult-onset, progressive neurodegenerative condition characterized by chorea, mental illness, and a decline in cognitive function. The mean age of onset is 35 to 44 years, with an average survival of 15 years after symptom onset (Haigh et al. 2004). HD is caused by a specific genetic mutation, discovered in the mid-1990s, which is inherited in an autosomal dominant fashion. Currently there is no cure, and symptoms can be difficult to manage. Prior to the mid-1990s, diagnosis was made from clinical criteria. There was no mechanism to identify, before symptom onset, which persons from at-risk families had in fact inherited the HD mutation and would eventually develop symptoms. However, once the genetic mutation responsible for causing HD was identified, a genetic test became available that could identify people with an HD-causing gene mutation many years prior to onset of symptoms. Given the fact that there was no cure for HD and recognizing that even the best medical management often could not eliminate symptoms, significant concern about the psychosocial ramifications of presymptomatic testing arose.

In an effort to protect the interests of at-risk individuals, the Huntington's Disease Society of America (HDSA) developed guidelines to assist healthcare professionals optimally manage the presymptomatic genetic testing process. HDSA recommends that the testing process be offered by organized, multidisciplinary HD testing centers incorporating the following components:

- Initial phone consultation that includes a prescreen interview with the at-risk individual
- Three pre-test appointments (often grouped into one visit in a specialty clinic), including
    - genetic counseling
    - neurological evaluation
    - psychological evaluation
- An appointment for the blood draw
- A genetic counseling session for face-to-face disclosure of results
- Post-test counseling sessions over a 2-year period

(Adapted from Huntington's Disease Society of America 1994)

MEET JASON

Clinical Example

Jason is a healthy, 20-year-old male who is currently attending a college that is 7 hours away from home. Jason's father calls to tell him that he has just learned his older brother, Brad (Jason's paternal uncle), has a genetic disorder called Huntington disease. Both the family and Brad's physician

now suspect that Jason's paternal grandmother died from the same condition. Jason's father explains that HD runs in families and that both he and Jason are at risk to inherit it. Jason's father suggests that he may want to talk to a healthcare professional near the college to learn more about HD and a genetic test he read about.

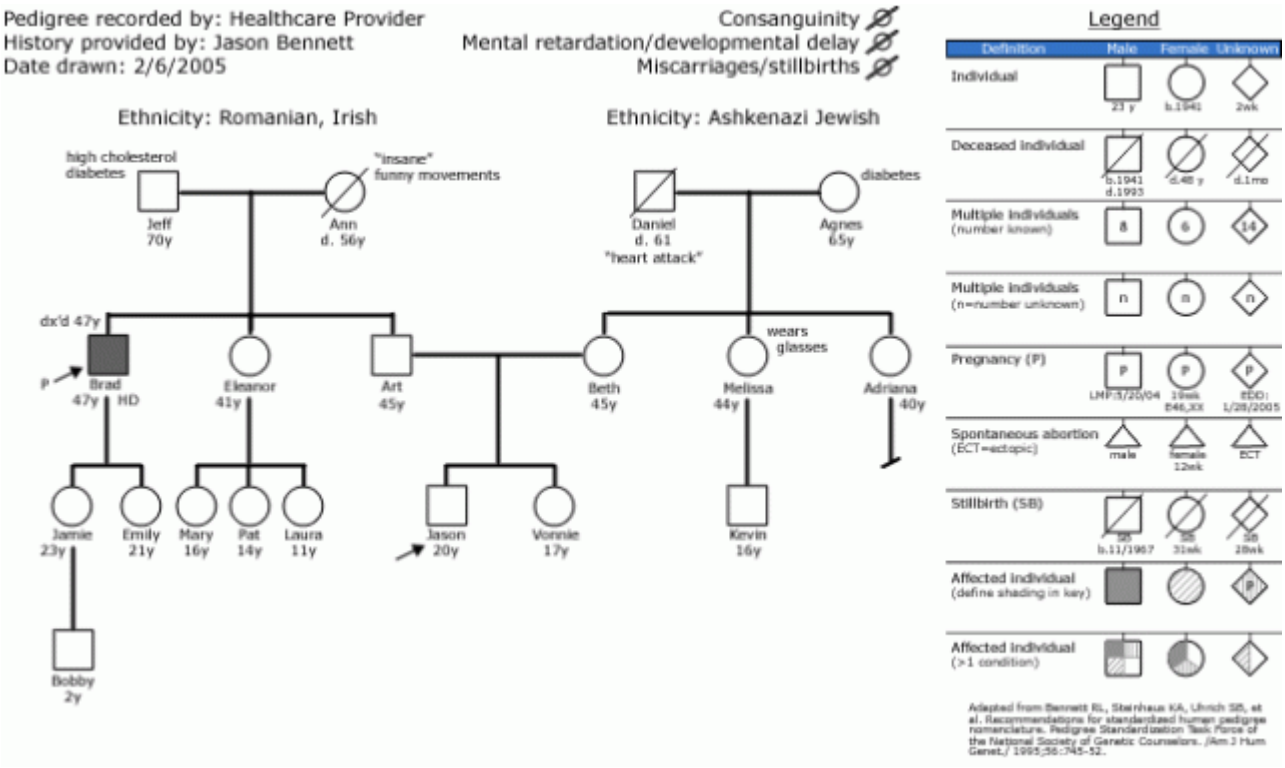
Jason has vague memories of his grandmother. She died in her mid-50s, when Jason was just 6 years old. Jason remembers going to visit her in a nursing home shortly before she died. She was strapped into a wheelchair and was making strange, jerky movements with her arms, legs, and face. Those movements were frightening to him as a child. He also remembers that his grandmother couldn't talk; and she had trouble swallowing, so she drooled. Jason has heard stories all of his life about how grandma went "insane." He recalls that she had to be put in an institution because she was having violent outbursts. Jason's grandfather was afraid that she would harm their children who still lived at home. Sometimes, when Jason's father's side of the family gets together, they jokingly recall odd things that his grandmother did. Family members describe their earliest childhood memories of his grandma repeatedly losing her keys and burning everything she cooked. Sometimes the stories aren't funny, recounting periods when she wouldn't come out of her room for days or times when she would snap at them. Jason never really knew what was wrong with his grandmother. He just accepted that she had "gone insane" for some unknown reason.

INFORMATION GATHERING

When Jason first seeks medical advice from you, all he knows about HD comes from his memories and the stories of his "crazy grandmother." He is terrified that he will follow a similar course. After Jason hurriedly describes the news his father called him with, the first question out of his mouth is, "Am I going to get it?"

You spend a few moments trying to put Jason at ease. Then, to help answer his question, you create a detailed 3-generation pedigree recording the medical history of Jason's family. Once complete, it appears that Jason's father's mother had symptoms consistent with HD. Her husband, Jeff, is currently 70 years old. He reportedly has high cholesterol and diabetes but no symptoms of HD. Jason does not have any medical information about his grandmother's family. Because she died when he was so young, he has not ever been in contact with them. Jason tells you that his dad has one sister, Eleanor, and one brother, Brad. Eleanor is 41 years old and reportedly showing no symptoms consistent with HD. Jason's description of his grandmother and uncle are consistent with a diagnosis of HD. However, you explain to Jason that you would like to confirm the diagnosis. You ask Jason to contact his father to see if Brad will release his medical records documenting the diagnosis of HD or, preferably, DNA mutation analysis results to you.

Jason's Family History



RISK ASSESSMENT

HD is inherited in an autosomal dominant manner, making risk assessment relatively straightforward. Assuming for the moment that Jason's grandmother did in fact have HD, Jason's father has a 50% chance to have inherited his mother's HD mutation. If Jason's father did inherit the mutation, Jason would then have a 50% chance to have inherited that mutation from his father. Thus, without first clarifying Jason's father's genetic status, Jason's risk to develop HD is 25% (1/2 x 1/2 = 1/4, or 25%).

Jason's risk to have inherited an HD-causing gene can be reduced from the 25% risk based solely on the autosomal dominant pattern of inheritance. This reduction is possible because the mean age of onset for HD symptoms ranges from 35 to 44 years. The fact that Jason's father is 45 years old and has no symptoms reduces the chance that he has a mutation in the HD gene. While Jason's father's risk at birth for inheriting the HD mutation was 50%, available data suggest that an asymptomatic 45-year-old actually has a 37.8% chance of having an HD mutation (Haigh et al. 2004), which would drop Jason's risk to 18.9% (37.8/100 x 1/2 = 37.8/200, or 18.9%). Should Jason's father remain asymptomatic at 55 years of age, his father's risk to have an HD mutation would drop further to 24.8% (Haigh et al. 2004), and Jason's risk would drop to 12.4%.

Below are the probabilities that an asymptomatic individual with an *a priori* risk of 50% for HD will test positive at a given age for an HD mutation.

Age at Testing	Probability of an HD Mutation
20 years	49.6%
25 years	49%

30 years	47.6%
35 years	45.5%
40 years	42.5%
45 years	37.8%
50 years	31.5%
55 years	24.8%
60 years	22.1%
65 years	12.8%
70 years	6.2%

(Adapted from Harper and Newcombe 1992)

COUNSELING

After your assessment of Jason's risk, you spend the rest of the appointment educating him about HD and the chance that he inherited an HD-causing mutation. During counseling, Jason learns that HD is a progressive neurodegenerative disorder characterized by chorea (involuntary movements), dementia, and psychiatric disturbances. The mean age of onset is 35 to 44 years, with an average survival of 15 years after symptom onset (Haigh et al., 2004). The timing and course of HD varies considerably between affected individuals, even within the same family. There are currently no effective preventive strategies, and treatment is primarily symptomatic.

You also discuss with Jason the availability of presymptomatic testing and explain the protocol suggested by the HDSA. Genetic testing is appropriate to offer to patients 18 years or older when a diagnosis of HD has been made in a family member. The presence of an HD disease-causing mutation in an affected family member should be confirmed, whenever possible, prior to testing unaffected family members. Jason's uncle likely had confirmatory HD genetic testing and hopefully will share those results with Jason, if Jason is interested in pursuing testing himself. Once the presence of a mutation is established in an affected family member, HD mutation analysis is technically straightforward and reliable. The presence of an HD-causing gene can be presymptomatically confirmed when there are greater than 36 CAG trinucleotide repeats at a specific site in the huntingtin gene.

While the genetic test for HD is technically clear-cut, the decision to pursue or decline testing is anything but straightforward. There is an extensive body of literature related to how and why individuals choose to accept or decline presymptomatic genetic testing and the impact that those decisions have over the short and long term. Some of the most commonly cited reasons used in deciding whether or not to proceed with presymptomatic genetic testing were determined by Evers-Kiebooms and Decruyenaere (1998). Those reasons are summarized below.

Benefits

- Desire for certainty or relief from uncertainty
- Make informed reproductive decisions
- Inform children about their risks
- Make practical life decisions such as financial

Barriers

- Perceived inability to cope with abnormal result
- Feeling that a test result does not dictate important decisions

and employment arrangements	• Not knowing results in more happiness than a known abnormal result • Lack of treatment options, if abnormal • Concern about reaction of family members • The extensive pre-test counseling protocol itself

While meeting with you, Jason is encouraged to explore what role being at risk for Huntington disease plays in his life. How is he coping with the new knowledge that he is at risk? What does he envision life as being like if he pursues testing and is found to have an HD mutation? or if he is found to be negative? or if he doesn't pursue testing at all? These are obviously complex questions that require time and serious consideration. The decision to pursue testing should be well thought out. Jason is relatively young and may want to delay deciding about testing until he feels a more pressing need, such as considering marriage or having children. On the other hand, he is currently in college and may feel that he will make career decisions based on the results and, therefore, needs to know now. Or he may feel he simply can't live with the uncertainty.

One additional question remaining for Jason to consider is what will happen if his father declines genetic testing but Jason decides to do so and is found to have a mutation? If Jason is positive, his father will, in effect, be presymptomatically diagnosed. While such a scenario would not prevent Jason from being tested, it does require some careful consideration and thoughtful discussion.

You end this first appointment with Jason by reminding him of the importance of confirming the diagnosis in his uncle, Brad. You also reinforce the fact that the information discussed during the appointment is only applicable if Brad does in fact have HD. Jason seems a little overwhelmed but tells you that he feels better now that he has learned about HD and understands how it runs through families. He says he definitely wants to be tested. He thanks you for the brochures and Web addresses you provided him. As he leaves, he tells you that he will call his father right away to work on getting documentation from Brad.

ONE WEEK LATER

One week later, you find in your mail a letter from Brad's neurologist confirming the diagnosis of HD, along with the results of Brad's genetic testing. As soon as you finish seeing previously scheduled patients, you call Jason. Jason was aware that Brad was having the documents mailed. He says that he is planning to talk to his family about the possibility of pursuing presymptomatic testing when he goes home for Thanksgiving in 2 weeks. Jason says now that he has had a little bit of time to think about it, it makes more sense to him to see if his father wants to pursue testing first. He also says that if his father tests positive, he (Jason) will probably want to follow through with presymptomatic testing but that he wants to be comfortable with his decision. He says the earliest he would start the process would be next summer in an effort to minimize the impact on school.

FOLLOW-UP

Four Months Later

You call Jason to check on him. Jason excitedly says that his father received his genetic test results last month and that his father does not have the HD mutation documented in Brad. Jason says that a huge source of worry has been lifted off of his shoulders. He thanks you for checking on him and for taking time to explain everything to him during his appointment. He says that understanding what was going on made him feel less anxious.

SOME FACTS TO CONSIDER

For At-Risk Individuals Who Pursue Testing

When an individual decides to pursue presymptomatic testing for HD, it is recommended that he/she receive genetic counseling, be examined by a neurologist, and meet with a psychologist. Many centers offering presymptomatic HD testing work with patients to coordinate these services into one visit. Once they have had time to digest the information and fully think through the ways test results might impact life, patients who elect to undergo testing will schedule a second appointment to have blood drawn. Usually an appointment for result disclosure is scheduled (2-3 weeks later) on the same day that blood is drawn for testing. Results are disclosed face-to-face. Patients should always be reassured that at any point during the protocol, even during the result disclosure appointment (but prior to actually receiving the result), they have the option to withdraw from the process, leaving their genetic status unknown.

Most studies suggest that, while patients receiving results indicating the presence of an HD-causing mutation have greater short-term psychological distress (as determined by standard psychological measures) than those who receive normal results, both groups of individuals adjust well over the long term. Adjustment to results seems to be impacted as much by psychological preparation prior to testing as by the result itself (Meiser and Dunn 2001). This finding supports the significance placed on the psychological evaluation during the pre-testing protocol. For patients with evidence of significant depression or hopelessness prior to testing, referral for ongoing psychological care should be considered before proceeding with testing.

Over 25% of individuals clinically affected with HD attempt suicide at least once, and 5.7% of deaths among affected individuals are due to suicide (Paulsen et al. 2005). While it is unclear how this suicide rate translates (if at all) to individuals determined by presymptomatic testing to have an HD-causing mutation, it raises obvious concern for healthcare providers involved with the testing process. Care must be taken to recognize patients at increased risk for suicide or for other poor outcomes.

A somewhat surprising finding is that a proportion of individuals who receive normal results (no HD-causing mutation present) have considerable difficulty coping. Such individuals report feelings of regret for having made irreversible life decisions based on their at-risk status or for having unrealistic expectations of the positive impact that a normal result would have in their lives (Huggins et al. 1992).

Another phenomenon, "survivor guilt," is also reported among patients receiving normal presymptomatic test results, particularly among those with family members who are currently affected or remain at risk. To help people adapt to presymptomatic test results, ongoing education and assessment is helpful. The HD testing protocol recommends post-test counseling over a period of 2 years for all tested individuals.

For At-Risk Individuals Who Decline Testing

For at-risk individuals who decide against or delay the genetic testing decision, reassurance that the option remains available to them at any point in the future should be provided. Wiggins et al. (1992) found that at-risk individuals who did not pursue genetic testing (available only by linkage analysis at the time of the study) had evidence of higher depression and lower well-being (as determined by standard scales) than patients who elected to proceed with presymptomatic testing, regardless of the actual test result. This study underscores the need for ongoing support of individuals who choose not to test.

All individuals either at risk or affected with HD should be provided written educational materials and referral to an appropriate local or national HD support group (such as *Huntington's Disease Society of America*).

SUMMARY AND KEY POINTS

- Genetic counseling is an educational communication process.
- During genetic counseling appointments, patient risks are ascertained through the creation of a 3-generation family history (or genetic pedigree). Ideally, reported diagnoses are confirmed by medical record examination.
- Risk and educational information is presented in a nondirective or value-neutral format so patients can utilize information in a way that is consistent with their personal values, beliefs, and life circumstances.
- All appropriate healthcare options should be presented to patients, including a discussion about any available genetic tests. Each genetic test should be discussed, along with its benefits and limitations.
- Follow-up counseling is offered to patients needing additional time to process information, make decisions, and work through emotions related to the genetic counseling process.
- Patients or families at risk for or diagnosed with genetic conditions are appropriate candidates for genetic counseling.
- Genetic counseling can be provided by any healthcare practitioner. Traditionally, master's-level genetic counselors, clinical geneticists, and nurses with specialized genetic training have offered counseling services. Today, genetic counseling is also provided by specialty clinics and primary care offices. As genetic knowledge and the number of genetic tests continue to grow, primary care physicians will increasingly encounter patients for whom genetic counseling is appropriate.
- *GeneTests* and *OMIM* are two invaluable resources created to help providers locate information about genetic disease, genetics professionals, and genetic testing.

RESOURCES AVAILABLE THROUGH THIS MODULE:

- American College of Medical Genetics
  The American College of Medical Genetics (ACMG) provides education, resources and a voice for the medical genetics profession. To make genetic services available to and improve the health of the public, the ACMG promotes the development and implementation of methods to diagnose, treat and prevent genetic diseases (From their Website). The website Includes information on resources, practice guidelines, and policy statements.
- American Society of Human Genetics
  The American Society of Human Genetics (ASHG), founded in 1948, is the primary professional membership organization for human genetics specialists worldwide. The Society's nearly 8,000 members include researchers, academicians, clinicians, laboratory practice professionals, genetic counselors, nurses and others who have a special interest in the field of human genetics. It aims to: Share research results at annual meetings and in The American Journal of Human Genetics, Advance genetic research by advocating for research support, Enhance genetics education by preparing future professionals and informing the public, and Promote genetic services and support responsible social and scientific policies (From their Website).
- Cancer Genetics Risk Assessment and Counseling
  The NCI has a nice discussion of the genetic counseling process tailored to the needs of patients at risk for or diagnosed with hereditary cancers.
- Definition of Genetic Counseling
  MedicineNet.com provides a general discussion about genetic counseling, including the psychosocial component of the process.
- GeneTests
  The GeneTests website offers an outstanding series of expert-authored GeneReviews that provide important information for clinicians to know about diagnosis, natural history, and genetic testing for genetic conditions. GeneTests.org also maintains databases of genetic testing laboratories and medical genetics clinics. There is no cost to use this website.
- Genetests Clinic Directory
  The Clinic Directory is a voluntary listing of US and international genetics clinics providing genetic evaluation and genetic counseling. It lists services, provides appointment contact information and links to clinic Websites, and information about clinic staff certification and credentials (From their Website).
- Genetic Alliance
  Genetic Alliance is a coalition of genetic advocacy organizations, health professionals, and health companies. The site contains a comprehensive, searchable directory of support and advocacy organizations. It also acts as a network of "stakeholders in the genetics community" to define policy, promote research, and protect the interests of its members.
- Genetics Education Center
  This website is for educators interested in human genetics. It contains many links about The Human Genome Project, Genetic Education Resources, Networking, and Genetic Programs/Resources.
- Huntington's Disease Society of America

The Huntington's Disease Society of America (HDSA) has a national office that produces and distributes, free of charge, publications and informational materials on Huntingon Disease (HD) and maintains a toll-free information hotline to assist physicians, patients and family members. Through 12 HDSA regions, 38 volunteer-based chapters and affiliates, 200+ support groups, they reach out across the nation to offer HD patients and their families guidance, encouragement, resource information and leadership opportunities at HDSA events, meetings and seminars (From their Website).

- Huntington Disease
  This web page provides a summary of the characteristics of Huntington Disease, as well information on the genetic testing used to diagnose the disease.
- Index of Rare Diseases
  This is the list of diseases currently covered in the Rare Disease Database (From their Website). With every disease, there is a link that provides a general discussion on the disorder, as well as a list of organizations and resources.
- Informed Consent during Genetic Testing
  Obtaining informed consent from patients is a vital part of the genetic testing protocol. Written documentation of informed consent is ideal. This page provides a list of what is generally included in informed consent.
- International Society of Nurses in Genetics
  The International Society of Nurses in Genetics (ISONG) is a global nursing specialty organization dedicated to fostering the scientific and professional growth of nurses in human genetics and genomics worldwide. Its mission is to foster the scientific, professional, and personal development of members in the management of genomic information. The website encourages users to incorporate new knowlege about human genetics into their practice, education and research activites. It contains documents prepared by members of ISONG as well as links to nursing and genetic sites which we find informative and helpful (From their Website).
- National Society of Genetic Counselors
  The National Society of Genetic Counselors is the professional membership association for the genetic counseling profession.
- Online Mendelian Inheritance In Man (OMIM)
  OMIM is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 12,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources. OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions (From their Website).
- Promoting Safe and Effective Genetic Testing in the US
  This is the final genetic testing report of the Task Force on Genetic Testing; the Task Force highlights principles and provides recommendations regarding genetic testing. The report includes chapters on: Ensuring the Safety and Effectiveness of New Genetic Tests, Ensuring

the Quality of Laboratories Performing Genetic Tests, Improving Providers' Understanding of Genetic Testing, and Genetic Testing for Rare Inherited Disorders.

- Recommendations for standardized human pedigree nomenclature (Abstract)
  Presents the recommendations of the Pedigree Standardization Task Force (PSTF) for standard human pedigree nomenclature and symbols. Full article is available.
- Surgeon General's Family Health History Initiative
  The Surgeon General's Family Health History Initiative is the national public health campaign to encourage all American families to learn more about their family health history. Moreover, it helps focus attention on the importance of family history since certain health conditions can be inherited and provides a "computerized tool to help make it fun and easy for anyone to create a sophisticated portrait of their family's health."
- The Family History Public Health Initiative
  A gateway to information and resources from the CDC for public health professionals about the potential for using family history as a tool for improving health and preventing or delaying the onset of common, chronic diseases.

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