GENETIC SERVICES AND THE PRIMARY CARE PROVIDER

Goal:
Take an active and appropriate role in the provision of genetic services to primary care patients.

After completing this activity participants will be able to:
- Use the personal and family history to recognize candidates for genetic services
- Select appropriate management strategies for patients with significant family histories
- Identify genetic testing situations that require genetic counseling and informed consent
- Describe the basic goals of genetic counseling
- Identify clinical genetics 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.

DEVELOPMENT OF THE FIELD OF MEDICAL GENETICS

History of the Field
Medical genetics is a relatively new field of medicine. While conditions have been recognized to run in families since Biblical times and although Gregor Mendel did his famous pea experiments in the mid-1800s, the first medical genetics clinics did not open until the 1950s (Harper 2008). Extraordinary advances have been made since that time. These include determining the correct number of human chromosomes, describing the structure of DNA, achieving the ability to diagnose conditions prenatally, and sequencing the human genome. All of these advances and numerous others have provided new tools to the field of medical genetics.

Throughout the history of medical genetics, understanding the role of hereditary factors in disease development has often significantly preceded the ability to offer effective management strategies (see Figure 1). Initially, medical geneticists focused on the delineation and diagnosis of relatively rare genetic disorders and birth defects. Recurrence risk estimation for family members was largely based on empiric data, with little more to offer than education and support (Walker 1998). As scientific knowledge about the genetic basis of inherited disorders improved, genetic tests were developed that enabled confirmation of clinical diagnoses. It also extended the application of medical genetics from those personally experiencing genetic disorders to broader applications, such as carrier screening for at-risk family members, population-based carrier screening programs, and prenatal diagnosis of chromosome abnormalities and many single-gene genetic conditions.

Figure 1. The Integration of Genetics Information Into Clinical Practice

CURRENT IMPACT OF THE FIELD OF MEDICAL GENETICS

Impact on Healthcare

Currently, the applications of new genetic technologies to the prediction and management of common disorders are blossoming. Nine of the 10 leading causes of death in the United States (excluding injury) have a genetic component (Guttmacher 2005). In addition, a significant percentage of morbidity among both children and adults is attributable to recognized genetic factors (see box below).

Public Health Impact of Genetics

Adult Medicine

- 12% of adult hospital admissions are for genetic causes (Emery and Rimoin 1990).
- 15% of all cancers have an inherited susceptibility (Schneider 1994).
- 10% of adult chronic diseases (heart, diabetes, arthritis) have a significant genetic component (Weatherall 1985).

Pediatrics

- 6% of all births result in congenital malformations (Christianson, Howson, & Modell 2006).
- 20% of all infant deaths are due to genetic disorders (Mathews & MacDorman 2012).
- Approximately 11.1% of pediatric hospital admissions are for children with genetic disorders and 18.5% are children with other congenital malformations (Scriver et al. 1973).

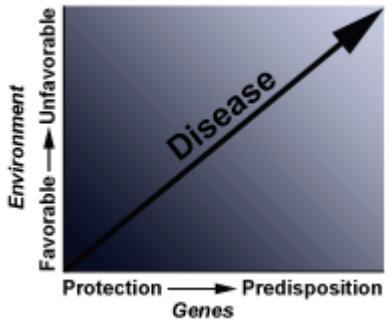
(Adapted from University of Kansas Medical Center 1995-2009)

EVOLUTION OF THE FIELD OF MEDICAL GENETICS

Direction of the Field

The amount of information being published about genetic variants found to predispose for or protect against common diseases is staggering. How these genetic variants interact with each other and an individual's environment to cause disease will be increasingly elucidated (see Figure 2).

Figure 2. Interaction of Genetics and Environment to Produce Disease



(Adapted from Scheuner and Yoon 2005)

Drs. Collins and McKusick state that "By the year 2010, it is expected that predictive genetic tests will be available for as many as a dozen common conditions, allowing individuals who wish to know this information to learn their susceptibilities and to take steps to reduce those risks for which interventions are or will be available" (Collins and McKusick 2001).

SHIFT FROM SPECIALIST TO PRIMARY CARE

As genetic information becomes an increasingly integral part of medical practice, primary care providers will be best poised to manage this integration for patients, for a number of reasons (Scott & Trotter 2013). Two significant reasons are the following:

- **A limited number of genetics professionals:** There are currently about 2,000 practicing genetic counselors in the United States and, while more difficult to quantify, there are likely even fewer clinical geneticists and genetic nurses. Thus, clinical genetics resources are expected to be deficient to meet the growing need for genetic services, especially in rural communities. Genetics professional resources will need to be used wisely.
- **The close bond between primary care physicians and patients:** Primary care providers have the distinct advantage of caring for patients (and often their family members) over an extended period of time. This longitudinal relationship increases the likelihood that a significant personal or family history will be recognized and allows the provider to offer and manage genetic services within the patient's familial, cultural, and psychosocial experiences.

DEFINING ROLES IN THE PROVISION OF GENETIC SERVICES

Korf (2002) offers a framework for defining the role of various clinicians (primary care, specialists, and medical geneticists) as the provision of genetic services continues to evolve (see Table 1 below). The term **genetic services** usually refers to the services offered by genetics professionals (genetic evaluation, testing, and/or counseling). However, nongenetics healthcare professionals may also choose to offer those genetics services that they are comfortable with providing.

The role of primary care and specialist clinicians will likely not change significantly for those individuals suspected to be affected with or at risk for relatively rare single-gene and chromosomal disorders (i.e., the more traditional genetic disorders such as cystic fibrosis, metabolic disorders, etc.). The major shift will likely occur in the management of common diseases. For many common conditions, there is a small subset of individuals who have a very strong genetic component to their disease, for which there may be a single gene of large effect (e.g., hereditary nonpolyposis colorectal cancer or familial hypercholesterolemia). The vast majority of those with common conditions, however, exhibit multifactorial inheritance, where there may be multiple genes involved (each with a small effect) that interact with one's environment to cause disease (e.g., diabetes, psychiatric disorders). Clinical geneticists will play a more prominent role when common diseases are likely to be

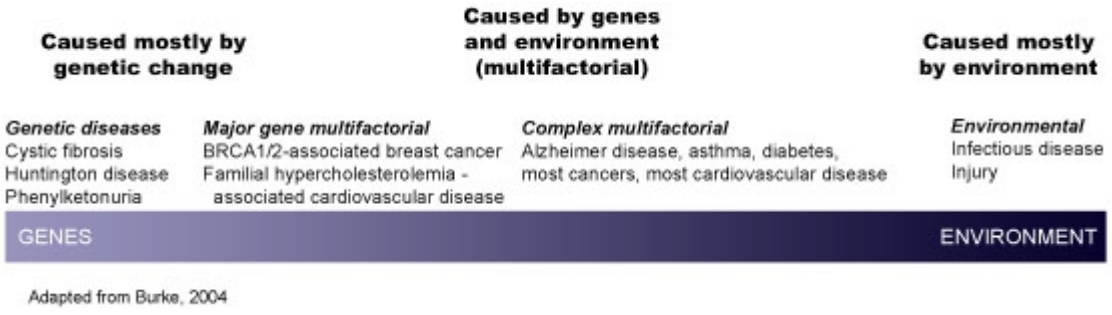
associated primarily with mutations in a single gene compared to when they are multifactorial conditions.

Table 1. The Role of Clinicians in 3 Different Genetic Service Models

Primary Care	Specialist	Medical Geneticist	
Single Gene or Chromosomal	- Recognize signs, symptoms, risk factors - Make referral - Support family - Provide longitudinal care in collaboration with specialists	- Manage system-specific problems	- Diagnose - Counsel patients/families - Provide longitudinal care in collaboration with primary care
Major Gene Multifactorial	- Recognize signs, symptoms, risk factors, and role of family history - Arrange testing and referral to specialists as needed - Provide longitudinal care	- Diagnose and manage system-specific problems	- Advise on interpretation of test results - Provide genetic counseling - Evaluate complex cases (e.g., inherited multisystem disorders that present with conditions such as cancer)
Complex Multifactorial	- Use personal/family history and genetic tests to guide prevention and treatment	- Use genetic tests to guide prevention and treatment	- Provide reservoir of knowledge and management of complex cases

(Adapted from Korf 2002)

Figure 3. Examples of Conditions With Single-Gene and Multifactorial Etiologies



Adapted from Burke, 2004

THE ROLE OF THE PRIMARY CARE PROVIDER

Primary care providers do not have to become experts in genetics to begin to provide genetic services. They simply have to do the following:

- Recognize when genetic factors may be play a role in disease development -- a process that Dr. Alan Guttmacher, of the National Human Genome Research Institute, calls "thinking genetically"
- Be familiar with available management strategies
- Know how to access genetics resources

RECOGNIZING PATIENTS WHO MAY BENEFIT FROM GENETIC SERVICES

The family history has long been the most useful and cost-effective "genetic test" (Bennett 2004). Most primary care providers obtain some family history from their patients; however, studies have shown that many individuals with a significant genetic risk are not identified by primary care providers (Frezzo et al. 2003; Meschede et al. 2000; Scheuner et al. 1997). When histories are collected in the primary care setting, they may not include enough family members, they may lack sufficient clinical information about affected relatives, or they may not get updated regularly.

There are many barriers to implementing a thorough family history (often defined as at least 3 generations) in primary care – time being perhaps the most difficult obstacle to overcome. Recognizing this constraint, tools to help elicit family history information directly from the patient are being developed and evaluated; some tools will even provide limited risk categorization and management decision support for a specific group of diseases (CDC 2004).

These sophisticated tools are not yet available, but the US Surgeon General's Office released *My Family Health Portrait* in 2004 as part of its Family History Initiative. It is a downloadable computer program available to the public that creates a pedigree through a brief interview process, and it includes some of the most commonly needed clinical information about affected relatives. A paper version is also available for those without a computer and Internet access. While not meant to replace the primary care family history, it may serve as a basis for determining who needs a more extensive history interview.

Whether a primary care physician uses a screening form, has a patient start or update his or her family history prior to the visit using a program or form, or obtains a complete family history in person, the provider must know how to recognize red flags. Generally speaking, individuals with findings that are uncommon or who have unusual presentations of common disorders should cause a healthcare provider to consider a genetic etiology. The Genetics and Primary Care Faculty Development Initiative developed 2 mnemonics to help providers successfully integrate family history into practice. The first, SCREEN (see box below), was developed primarily by Dr. Caryl Heaton as a reminder of the important questions to ask during the family history interview (Porter 2005).

SCREEN: Questions to ask during the history interview

S C	**S**ome **C**oncern: Do you have any concerns about diseases or conditions that seem to run in your family?
R	**R**eproduction: Have there been any problems with pregnancy, infertility, or birth defects in your family?
E	**E**arly disease, death, or disability: Have any family members died or become sick at an early age?
E	**E**thnicity: What is your ethnic background -- where did your ancestors come from?
N	**N**ongenetic or **N**ot necessarily genetic: Are there any other traits or nonmedical conditions that seem to run in your family?

(Adapted from Porter 2005)

FamilyGENES: Consider genetic causes in the differential diagnosis

Family	Positive **Family** history
G	**G**roups of anomalies
E	**E**arly or **E**xtreme presentations of common disease
N	**N**eurodevelopmental or **N**eurodegenerative conditions
E	**E**xceptional or unusual pathology
S	**S**urprising laboratory values

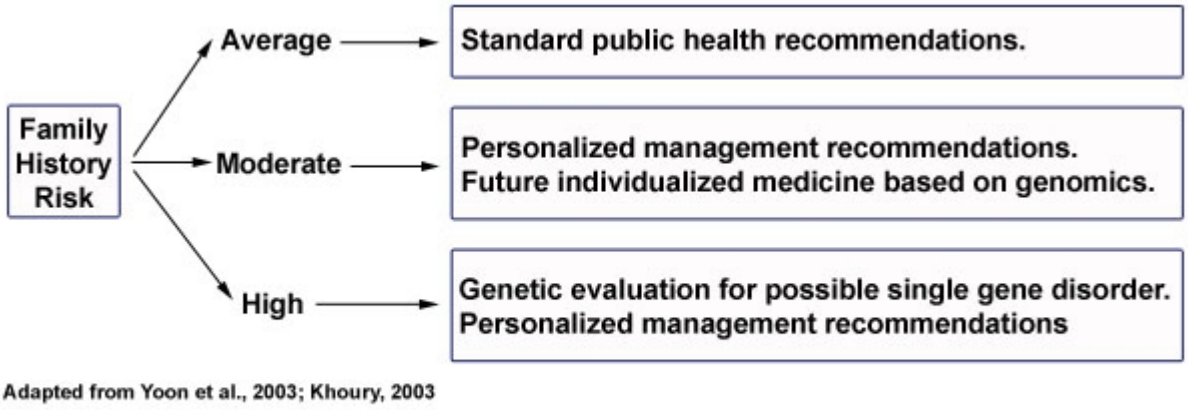
(Adapted from Whelan et al. 2004)

IDENTIFY MANAGEMENT OPTIONS

Management Options

Once a personal or family history is found to be significant, the primary care provider must decide if the history demands an altered management strategy. Options may include increased surveillance, risk-reducing behaviors, some form of testing, genetics referral, and/or other interventions. Virtually every traditional single-gene disorder and chromosome abnormality requires altered management in the clinically affected individual and often has management implications for at-risk family members. Referral to clinical genetics and possibly other specialists are appropriate management options.

Adjust General Population Screening and Management Strategies for Common Diseases



Adapted from Yoon et al., 2003; Khoury, 2003

It is important to recognize those family histories with common conditions that seem suspicious for a single-gene etiology versus the more common multifactorial inheritance. In general, you should consider a single-gene etiology when the age of onset among affected family members is significantly earlier than usual, when there are 2 or more affected relatives, and/or when there is an unusual presentation. Family members of individuals with single-gene explanations for their disease are generally at much greater risk than those with a more typical positive family history. As a result, management options may be highly individualized once the etiology is recognized.

WHEN SHOULD I REFER PATIENTS TO A GENETICS PROFESSIONAL?

Any healthcare provider who is making a genetic diagnosis, assessing patient risk for a genetic condition, ordering genetic testing, and/or interpreting genetic test results is providing some level of genetic service. For the primary care provider, the decision is often how much genetic service to personally provide. At present, there are no broad-sweeping published standards to guide that decision, although some guidelines do exist for specific clinical circumstances (e.g., presymptomatic diagnosis of Huntington disease). Ultimately, referral decisions should take into account one's knowledge base, one's ability to adequately provide a complete genetic service, and the availability of genetics resources.

INDICATIONS FOR GENETIC SERVICES FOR ADULTS

In general, adults who either are at increased risk for a genetic condition or are suspected to have a genetic condition are candidates for genetic services. The box below contains a helpful list of indications that suggest genetic services are appropriate.

The application of genetic technologies to predictive testing for adult-onset conditions is a relatively recent frontier of genetic medicine. For conditions with high penetrance, genetic testing may provide

knowledge of an impending condition before symptoms are apparent. For other conditions, genetic test results may only indicate an increased predisposition toward disease -- onset of disease is not guaranteed. One of the most challenging aspects of adult genetics is identifying patients who are at greatest risk for developing disease with an underlying genetic component, so that reasonable management options may be offered. Healthcare options may include increased surveillance, prophylactic surgeries/interventions, lifestyle changes, and/or genetic testing. The ethical, legal, and psychosocial issues surrounding predictive testing are significant.

Common Indications for Genetic Services in Adult Medicine (Not Reproductive)

- Personal or family history of a known or suspected genetic condition or chromosomal abnormality that may present in adulthood or requires ongoing management into adulthood
- Personal or family history of common conditions with unusual presentations (early onset or more severe course, combined with other unusual findings)
- Personal or family history suspicious for hereditary cancer syndrome (early onset, clustering of cancers that may have the same etiology, multiple primary cancers in a single person, bilateral disease in paired organs, rare cancers or tumors)
- Ethnicity known to be associated with increased risk for specific genetic conditions (BRCA mutations more common in Ashkenazi Jewish individuals)
- Development of a degenerative disease
- Sudden premature death in an apparently healthy person

(Adapted from GeneTests 1993-2005)

INDICATIONS FOR GENETIC SERVICES FOR CHILDREN

Most birth defects and genetic conditions appear without warning in families that have no previous history of the disorder. Families coping with an affected child have a tremendous need for multidisciplinary care, education, advocacy, and support. Some of the most common indications for a pediatric genetics consultation are listed below.

Common Indications for Pediatric Genetic Consultation

- Abnormal newborn screening results
- One or more major malformations in any organ system
- Abnormalities in growth
- Mental retardation or developmental delay
- Blindness or deafness
- Presence of a known or suspected genetic disorder or chromosomal abnormality
- Family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality

(Adapted from GeneTests 1993-2005)

INDICATIONS FOR GENETIC SERVICES FOR EXPECTANT PARENTS

The chance of a condition occurring or recurring in one's children has long been a concern for anxious parents. In the early years of the field of medical genetics, when a significant reproductive risk was identified for a couple, usually the only option the couple had was to decide whether or not to have children. However, since amniocentesis has become widely available, at-risk families have a number of options for managing their reproductive lives. Depending on the clinical situation, patient options range from preimplantation genetic diagnosis to first-trimester nuchal translucency screening to *in utero* fetal surgery. The rapidly increasing number of options has steadily led to a greater need for reproductive genetic counseling. Some of the most common indications for reproductive genetic counseling are listed below.

Common Indications for Reproductive Genetic Services

- Maternal age of 35 years or older at delivery
- Abnormal results from a maternal serum screen (first- or second-trimester screening) or fetal ultrasound
- Personal or family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
- Ethnic background associated with a predisposition to certain genetic disorders (such as Ashkenazi Jewish)
- Abnormal carrier-screening result on routine population screening (such as cystic fibrosis)
- Exposure to a known or potential teratogen
- Maternal medical condition known or suspected to affect fetal development
- Two or more pregnancy losses (miscarriage or stillbirth)
- Close biological relationship of parents (consanguinity)
- Infertility, particularly when due to oligospermia or azoospermia, congenital absence of the vas deferens, or premature ovarian failure or when there are indications that a chromosome abnormality may be present

  (Adapted from GeneTests 1993-2005)

INTRODUCTION TO GENETIC SERVICES

Genetic services is a broad term that, for the purposes of this course, includes the following:

- **Genetic testing:** Examples include the following:
  - diagnostic testing
  - predictive testing
  - presymptomatic testing
  - carrier testing
  - prenatal testing
- **Genetic evaluation:** Examples include the following:
  - dysmorphology examination

    - pedigree review
    - medical record review
- **Genetic counseling:** Examples include the following:
    - risk assessment
    - nondirective patient education
    - facilitating patient decision making

GENETIC TESTING

Testing for genetic conditions may be accomplished through DNA-based testing, cytogenetics studies, or biochemical assays -- depending on the etiology of the condition. The indication for testing is generally for diagnostic, carrier-screening, prenatal diagnostic, or predictive purposes. A genetic test is usually only appropriate for use in very specific clinical circumstances. For instance, when testing for hereditary breast and ovarian cancer syndrome, there are four different clinically available BRCA tests: full-gene sequence analysis including five specific rearrangements, mutation analysis for 3 mutations common in Ashkenazi Jewish individuals, single-site mutation testing for known familial mutations, and BRCA rearrangement test. If the correct test is not selected, the results are likely to be meaningless for the patient (not to mention costly and potentially harmful). To determine if genetic testing is appropriate for a specific clinical circumstance, the following factors must be considered:

Adapted from Haddow and Palomaki, 2003

- Is a test available? on a clinical basis? only on a research basis?
- What is the validity of the test for the clinical circumstances (sensitivity, specificity, penetrance of an identified mutation, quality of the particular assay, etc.)?

- How useful will the results be in clinical management (guide appropriate interventions, define diagnosis or prognosis)?
- What are the ethical, legal, and psychosocial implications of pursuing or declining testing?

Many genetic testing laboratories employ genetic counselors, PhD-level geneticists, or other genetics specialists. These individuals can be great assets to the primary care provider who is considering ordering a genetic test. Generally speaking, testing is available for common single-gene or chromosomal conditions at major laboratories. Testing for less common genetic disorders usually occurs in academic or specialty laboratories. One extremely valuable resource to aid healthcare providers in locating laboratories offering genetic testing is *GeneTests*. This website contains the most comprehensive list of genetic tests available and is free to use.

The number of common conditions for which predictive genetic tests are available is increasing rapidly. Some of these are valid and useful in carefully selected circumstances (such as several cancer syndromes, hemochromatosis, or factor V Leiden-associated thromboembolism). Other studies are clinically available, but their validity and utility is generally poor (e.g., ApoE analysis for Alzheimer disease). Most concerning are the laboratories offering unfounded predictive genomic panels (often directly to the consumer) to assess risk for such outcomes as cardiovascular disease, osteoporosis, poor nutritional status, and mental health conditions.

CLINICAL GENETICS EVALUATIONS

Evaluations

Two main types of clinical genetics evaluation exist; they are described below.

- Evaluations performed by physician geneticists that include physical examinations
- Evaluations performed by a physician geneticist or other healthcare professional (genetic counselor or genetic nurse) that do not include physical examinations

The type of evaluation performed will depend on the indication for the evaluation. In general, when a differential diagnosis involving physically apparent hallmarks is being considered, a physical examination will be performed. For example, children born with birth defects (a cleft palate, heart defect, etc.) need to have a physical examination. In this situation, a very unique physical examination, a dysmorphology exam, will be performed. During the examination, body parts (head circumference, inner and outer canthal distance, ear size, etc.) are measured and then compared to accepted normal standards. The presence or absence of additional anomalies is also noted. After the examination, the physician will try to determine a diagnosis for the child.

When diagnoses being considered are either not physically apparent or occur in a relative of your patient, a physical examination may not be needed. Instead, a detailed family history, medical history, and record evaluation may be sufficient to assess potential risk for your patient. For example, if your patient is a 28-year-old pregnant woman of normal appearance and intelligence who raises concerns about her baby inheriting Down syndrome "since her cousin has it," a physical examination will be pointless. To best evaluate this woman's risk, medical records confirming a diagnosis in her cousin would be most helpful. If medical records are unavailable, chromosome analysis of your patient could be performed to confirm that she has a normal, female karyotype (46, XX).

Clinical Genetics Professionals: Physicians, Genetic Counselors, and Genetic Nurses

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, physician geneticists work in conjunction with genetic counselors and/or genetic nurses. Although each clinic is unique, physician geneticists usually review medical records, conduct physical exams, and attempt to determine or confirm a diagnosis. Often, the genetic counselors and nurses provide the bulk of information gathering, risk assessment, and counseling. Neither genetic counselors nor genetic nurses make diagnoses. Genetics clinics with physician geneticists are often located in large academic medical centers or outreach programs affiliated with such centers.

GENETIC COUNSELING

Many people in the genetics community consider genetic counseling to be the cornerstone of genetic services. The term genetic counseling was coined by Sheldon Reed in 1947 (Resta 2006). However, it wasn't until 1975 that the American Society of Human Genetics published the still-used definition and goals of genetic counseling (see the box below) (National Cancer Institute 2014).

Any clinician who enters into genetic counseling with a patient needs to be prepared to adequately address each goal. Even if you do not plan to personally provide genetic counseling, it is useful to be familiar with these goals so that you can prepare a patient for genetics referral and take an active role in the process. Each genetic counseling goal will be discussed in the remaining pages of this chapter. Patients who are appropriate candidates for genetic services will likely seek the advice of the healthcare provider that they know best, their primary care provider. Thus, even if they prefer to refer patients to genetics professionals, primary care physicians will likely field questions, be asked to provide opinions, and deal with the long-term implications of whatever the risk assessment or testing reveals.

Genetic Counseling Definition and Goals

Genetic counseling is a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained people to help the individual or family in the following ways:

- "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 which seems to them appropriate in view of their risk, their family goals, and their 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."

  (Quoted from National Cancer Institute 2014)

GENETIC COUNSELING: EDUCATION

"Comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management" (National Cancer Institute 2014).

Adequate education is the foundation for informed consent. In order for patients to make informed decisions about any testing or treatment option, they need to receive accurate and up-to-date information about the symptoms, natural history, and inheritance of the condition(s) for which they and/or their children are at risk. Patients must also understand all of their management options, including the personal, social, and financial implications of pursuing or declining each of these options. The following are websites that patients may find useful.

GENETIC COUNSELING: RISK ASSESSMENT AND COMMUNICATION

"Appreciate the way that heredity contributes to the disorder, and the risk of recurrence or occurrence" (Epstein et al. 1975).

"Understand the alternatives for dealing with the risk of recurrence or occurrence" (Epstein et al. 1975).

Risk assessment for the occurrence or recurrence of a condition may range from being quite straightforward (as when analyzing a pedigree for a Mendelian condition with complete penetrance) to being very complex (as when using multiple statistical models to assess risk for hereditary breast cancer). Identified risks should be presented to the patient along with the risks, benefits, and limitations of various management options. Risk assessment is useful only when the patient is able to understand the risks and benefits presented. Patients cannot make appropriate, informed decisions about risk-reducing behaviors, testing options, and other interventions if they do not understand the information provided.

Effectively communicating statistical information about risks and benefits is not specific to the field of medical genetics. Instead, it is a common challenge in all areas of medicine. The average patient has a limited capacity for understanding probabilities and numerical data (or **low numeracy**), even among patients with a high school education or a college degree (Reyna et al. 2009). The patient's concept of personal risk results from a combination of his or her understanding of the objective data in addition to the perceived burden of the outcome, its controllability, personal experience with the outcome, and preconceived notions of risk (Gates 2004; Vlek 1987).

A further complication to risk perception is that during genetic counseling interactions healthcare providers are often asking individuals to simultaneously comprehend multiple risks, comprehend complicated risk ranges, and to deal with a degree of uncertainty. Julian-Reynier et al. (2003)

reviewed risk communication strategies in the cancer genetics setting and found that the typical patient was provided risk information in about 6 different areas:

- The likelihood that a cancer gene mutation is running in the family
- The risk of having inherited the mutation (if one is present in the family) and the risk for transmitting that mutation to offspring
- The chance of detecting the cancer gene mutation through available testing (if a mutation is present)
- The risks of developing cancer if the mutation is present or **not** present
- The risks associated with various prevention, early detection, and other management options
- The risk of recurrent cancers or other health effects associated with a hereditary cancer syndrome

Comprehension and retention of all of this information is undoubtedly an overwhelming task for most patients. Thus, genetic counseling is often provided over a series of visits. Much attention must be paid to effectively communicating risk and benefit data and also assessing patient understanding.

GENETIC COUNSELING: FACILITATING DECISION MAKING

"Choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, and act in accordance with that decision" (Epstein et al. 1975).

The process of genetic counseling is generally described as **nondirective**, meaning "procedures aimed at promoting the autonomy and self-directedness of the client" (Kessler 1997). Medical genetics, as a specialty, evolved in the aftermath of World War II, at a time when the public was very aware of the dangers of misplaced goals in the name of eugenics. It is generally believed that public concern regarding the use of genetic information greatly influenced geneticists' commitment to patient autonomy. Geneticists adopted a nondirective counseling approach (Resta 1997b), particularly in matters related to reproduction. While primary prevention, gene therapy, and other interventions are the promise of the Human Genome Project, relatively few examples of highly effective management options currently exist for genetic conditions. As a result, the reality persists that genetic information is often sought out more often for personal than for medical reasons. Reasons may include family planning, financial decision making, preparation for the future, as well as many others.

The term ***nondirective*** has been contested as being an inappropriate description of the genetic counseling process and an unattainable goal. Several other terms have been suggested, including ***shared decision making*** and ***value-neutral counseling***. Regardless of the terminology used, for situations where there is no clear medical indication for testing and another management option is not available (e.g., prenatal screening and diagnosis, presymptomatic testing for an untreatable condition), genetic counseling should have the goal of empowering individuals through education and counseling to make genetic choices that are consistent with their personal and familial values, beliefs, and needs.

The genetic counseling goals do **not** prevent the provision of medical advice when there is a clear medical benefit to one course of action over another. For instance, the values of genetic counseling

would not support recommending genetic testing for hereditary nonpolyposis colorectal cancer to a person with a suggestive family history because the clinical utility is unclear; but appropriate colonoscopy screening would be strongly recommended.

Burke and colleagues (2001) provide a framework for assessing the appropriateness of nondirective counseling based on the type of genetic testing decision to be made. This categorization is based on the availability of effective intervention and the clinical validity of the test. They argue that when an effective, acceptable treatment exists, "healthcare providers may have an obligation to provide both testing to those eligible and the associated treatment to those who test positive." However, when an effective, acceptable treatment is **not** available, "the importance of nondirective counseling becomes paramount," and the focus should be on personal choice.

Effective, Acceptable Treatment Available?		
Clinical Validity:	**Yes**	**No**
High	**Examples:** Multiple endocrine neoplasia, familial adenomatous polyposis, phenylketonuria **Approach:** Test recommendation is appropriate (does not negate autonomy) **ELSI* focus:** Access to testing and treatment	**Examples:** Huntington disease, myotonic dystrophy **Approach:** Careful, nondirective pretest counseling promoting autonomous decision making **ELSI* focus:** Risk of adverse labeling, psychological distress, potential for discrimination
Low	**Examples:** Hemochromatosis, hypercholesterolemia **Approach:** Variable, depending on data related to medical utility **ELSI* focus:** Balance between adverse effects of labeling or treatment and potential for improved health outcome; requires good data	**Example:** ApoE genotyping for Alzheimer disease risk assessment **Approach:** Testing difficult to justify on medical or social grounds; appropriate to advise against testing **ELSI* focus:** Obligation to avoid harm related to unrealistic expectations of predictive value by patients; potential injustice of using resources to counsel about and implement testing of limited use

*ELSI = ethical, legal, and social implications)

(Adapted from Burke et al. 2001)

Value neutrality is often a difficult approach to adopt, particularly for healthcare providers who are accustomed to providing patients specific advice and direction. An important factor in successfully providing nondirective counseling is understanding one's own values, personal biases, and decision-making processes as well as recognizing that many forces shape decision making and that each

patient is unique. Self-awareness will help minimize the unconscious role these factors may play during counseling. Increased awareness of diversity may also promote respect for patient decisions that are inconsistent with one's own beliefs.

GENETIC COUNSELING: FOLLOW-UP

"Make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder" (Epstein et al. 1975).

Most patients report a significant amount of anxiety related to genetic decision making, the genetic testing process, and/or the birth of a child with a genetic condition. For most, these emotions subside through a normal process. However, healthcare providers need to recognize those patients who are experiencing ongoing, significant emotional distress and be prepared to make a referral to a local mental health professional -- preferably one experienced in dealing with the unique psychosocial issues related to these experiences -- for ongoing counseling.

Whether the outcome is the diagnosis of a genetic condition in a child, the presymptomatic detection of a disease-causing mutation, or simply the identification that an individual is at risk, ongoing education, advocacy, and support will be required.

There are a variety of advocacy and support services designed specifically for individuals with genetic concerns. In well-populated areas, particularly areas with a university hospital system, there are often local specialty clinics and support groups for the most common diagnoses (e.g., Down syndrome, sickle cell anemia, muscular dystrophy). In addition to local resources, the Internet hosts a wealth of patient-oriented information available to anyone with Internet access. Patient-oriented sites include patient/family advocacy informational websites, online discussion boards, and matching services for individuals with rare conditions. Because anyone may post information on the Internet, it is best to provide patients with specific, high-quality resources to help ensure that the information is accurate and appropriate. Some resources that are especially helpful are included in the box below.

SUMMARY AND KEY POINTS

As genetics information becomes a more integral part of medical practice, primary care providers will play an increasing role in managing genetic information. They will identify patients that should consider genetic services and will manage the testing and treatment process for many.

Obtain Family History Information: Obtaining a family history is the single most important step a primary care provider can take. As traditional gatekeepers to specialist care, primary care providers have the responsibility to recognize patients that will benefit from genetic services.

Determine Your Role: Your role in the management of a genetic disorder will depend on the level of risk revealed by the family history. The primary care provider must decide if a significant family history warrants an altered management strategy. Options might include the following:

- Increased surveillance
- Risk-reducing behaviors
- Some form of testing

- Genetics referral
- Other intervention

Define an Appropriate Management Strategy:

- Determine if the general population screening and management guidelines are altered based on the personal or family history.
- Determine if genetic testing is available and appropriate to the clinical circumstances.
- Determine if a genetics referral is indicated.

Be Familiar With the Genetic Counseling Process: Understanding the nature of the genetic counseling process will enable you to determine if you are able to adequately provide such services and/or prepare patients for a genetics referral.

RESOURCES AVAILABLE THROUGH THIS MODULE:

BRACAnalysis Technical Specifications

This article includes information on the four different clinically available BRCA tests for the testing of hereditary breast cancer syndrome: comprehensive full-gene sequence analysis including five specific rearrangements, single site testing for known familial mutations, multisite test, and the rearrangement test. It also includes information on the performance characteristics and interpretive criteria of the tests.

GeneTests.org

The GeneTests Web site is a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons. (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.

Genetic and Rare Diseases Information Center (GARD)

This center was established by the National Human Genome Research Institute and the Office of Rare Diseases. GARD includes specific information on: what is known about a genetic or rare disease, what research studies are being conducted, what genetic testing and genetic services are available, which advoacy groups to contact for a genetic or rare disease, and what has been written recently about a genetic or rare disease in medical journals (From their Website). The Information Center provides assistance to patients and families, health professionals, and other interested parties." It receives questions by phone, e-mail, fax, or mail and will respond in English or in Spanish.

Genetic Health

This is a website providing helpful information about general genetics, the genetic testing process, and ethical and legal issues related to genetic diagnosis, research participation, resources, and managing your risk. The site also has information describing a few adult-onset genetic conditions.

Genetics/Birth Defects Topics

This section of MedlinePlus has a list of about 100 genetic conditions and birth defects. For each condition listed, there are many links to reputable online resources for general disease information, current research, diagnostic testing, and support organizations.

Genetics Home Reference

The Genetics Home Reference website provides consumer-friendly information about the effects of genetic variations on human health, and is a guide to understanding genetic conditions (From their Website). It offers information about basic genetics, some genetic diseases and the underlying etiology of such diseases, and many helpful illustrations.

Genogram Template

An example of a general genogram with many symbols and their relationships to the patient; includes a key for common nomenclature. Genograms allow the visualization of hereditary patterns within a family.

Informed Consent

Informed consent allows a patient to make fully educated healthcare decisions.

Medical Family History Red Flags

A list of family history red flags

My Family Health Portrait

This program, from the US Surgeon General's Family History Initiative, enables people to answer a series of questions about the number, relationship, and medical histories of their family members. This information is then used to generate a pedigree that the patient can bring to his or her physician for review. The program is relatively simplistic (does not distinguish half siblings from full siblings, includes a limited number of relatives, no risk assessment, etc.), but the Surgeon General's office plans to continue developing the tool. It is simple for the patient to complete, requires only about 10 to 15 minutes (if all of the health information is readily available), and the information is kept on one's own computer, minimizing security concerns.

National Organization for Rare Disorders

The National Organization for Rare Disorders (NORD) is a unique federation of individuals and organizations working together to build a better world for people affected by rare diseases (From their Website). This organization maintains a database of consumer friendly reports for over 1,100 rare diseases (many of which have at least a genetic component). The site also has a searchable index of support and advocacy organizations.

Prevalence of Genetic Conditions/Birth Defects

Provides data on the prevalence of birth defects and various genetic conditions. Additionally, it includes information on international prevalence and includes resources on the lifetime cost of care for certain conditions.

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