Genetic Counseling

3

Christine E. Miller

Abstract

Genetic counseling is a relatively new profession that began four decades ago. Genetic counselors are master level-trained individuals who have specialized knowledge in medical genetics and counseling. About 83 % work directly with patients on a daily basis, while approximately 17 % work in laboratories or other non-patient contact areas. Genetic counselors are trained to be nondirective and advocate for patient autonomy and informed consent. The genetic counseling process involves drawing a family medical pedigree, reviewing medical records, performing risk assessments, explaining medical and scientific information, discussing disease management, treatment and surveillance options, reviewing testing options, and facilitating the decision-making process. Genetic counselors believe that patients will make the best choice for themselves if their decision is made in the context of their belief systems and past experiences. Genetic counseling positions and subspecialties are rapidly evolving to meet patients' needs. Given the speed of gene discovery, genetic counselors must work to keep abreast of knowledge of specific diseases, innovative testing methods, and new disease treatments and surveillance options.

Keywords

Genetic counseling • Genetics • Informed consent • Risks • Benefits • Screening • Testing • Patient • Family • Proband • Family history • Autosomal • Dominant • Recessive • X-linked • Carrier • Ethnicity

Genetic Counseling: The Discipline and the Provider

Genetic counseling is a relatively new healthcare profession rooted in a combination of medical genetics and counseling theory. The first program to train master-level genetic counselors was Sarah Lawrence College (New York) graduating its first class of eight students in 1971 [1]. Due to the increasing demand for genetic counseling services in the era of genomic medicine, by 2012, there were a total of 31 master-level genetic counseling training programs in the USA and over 2,500 practicing genetic counselors [2]. The American Board of Genetic Counseling (ABGC) provides accreditation for genetic counseling programs that meet specific educational criteria and it administers a national certification examination for genetic counselors. Thirteen states now regulate the profession of genetic counseling requiring genetic counselors to obtain licensure prior to providing this service to citizens of their state. Of the genetic counselors who provide direct patient counseling, approximately 32 % are employed in prenatal genetics, 22 % in cancer genetics, and 14 % in pediatric genetic settings. Thirty-two percent of genetic counselors have expanded into other subspecialties, including artificial reproductive technology (ART), neurology,

C.E. Miller, M.S., C.L.G.C. (✉)
ARUP Laboratories, 500 Chipeta Way, MS D-115,
Salt Lake City, UT 84108, USA
e-mail: MILLERC@aruplab.com

D.G.B. Leonard (ed.), *Molecular Pathology in Clinical Practice*, DOI 10.1007/978-3-319-19674-9_3

Table 3.1 Process of genetic counseling

Precounseling assessment	Construct medical pedigree, review pertinent medical records, and perform clinical assessment
Risk assessment	Calculate risk for occurrence using medical pedigree, test results, medical literature, and Bayesian analysis
Counsel patient, couple, or family	Explain medical and scientific information, discuss disease management, treatment and surveillance options, review various testing options, facilitate decision-making process and order appropriate genetic test(s)
Follow up	Summarize discussion in written form for referring healthcare provider and consultant(s), share information about support groups or patient-friendly information on the Internet, provide referrals to psychotherapists or family therapists, as necessary

psychiatry, cardiology, ophthalmology, and genomic and laboratory medicine. Approximately 9 % of genetic counselors work in diagnostic laboratories providing education and support to the patients and physicians who request help with genetic test selection and result interpretation [3]. Genetic counseling is a profession that actively responds to the changing landscape of medicine and matches the trends and needs in the healthcare marketplace.

Physicians with a subspecialty in genetics also provide genetic counseling services. Physician geneticists have a background in various disciplines such as obstetrics, pediatrics, and internal medicine have obtained specialty training in medical genetics and are board certified by the American Board of Medical Genetics (ABMG). As genomic medicine evolves into mainstream healthcare, all providers will need some genetic training to be able to order appropriate tests for their patients and understand the clinical significance of molecular and genomic test results.

Genetic counseling is a process whereby a genetic counselor (1) elicits a family history to construct a medical family pedigree, (2) obtains necessary records to confirm reported diagnoses, (3) determines the risk for occurrence or recurrence of a specific disorder(s), (4) communicates the natural history of the disorder(s) and risk for recurrence, and (5) explains screening, testing, or treatment options to the patient/family in a nondirective manner helping them make the best possible adjustment to the condition(s). Table 3.1 summarizes the key stages of the genetic counseling process. In 2007, the National Society of Genetic Counseling (NSGC) released an expanded scope of practice which included genetic counselors' ability to order genetic tests and perform clinical assessments in accordance with state and federal regulations [4]. The scope of practice will need continual reassessment to meet patients' needs in this rapidly evolving profession.

Clinical Genetic Counseling

Prenatal Setting

Women are referred for prenatal (and less commonly preconception) genetic counseling and prenatal diagnosis for numerous reasons including advanced maternal age (>34 years), abnormal screening test results, abnormal ultrasound findings, family history of an inherited condition, consanguinity, teratogen exposure, and multiple miscarriages or stillbirths. Although all women are at risk to have a child with a chromosome abnormality, this risk increases with maternal age. Thus, advanced maternal age remains one of the most common reasons a woman is referred for prenatal genetic counseling. In 2007, the American College of Obstetrics and Gynecology (ACOG) recommended all women be offered first- and second-trimester maternal serum screening (MSS) for Down syndrome, trisomy 18, and open neural tube defects [5]. ACOG stated that women of all ages should have the option of invasive diagnostic testing. This is a large departure from when prenatal diagnosis was mainly offered to women 35 years and older in the 1970s and early 1980s.

Amniocentesis remains the most common prenatal diagnostic technique. The procedure involves guiding a needle through the abdominal wall into the uterus and withdrawing 20 cc of amniotic fluid, which contains fetal cells, for cytogenetic and molecular analysis. Optimally, amniocentesis is performed under ultrasound guidance between 14 and 18 weeks of gestation, but can be performed safely throughout the remainder of pregnancy as well. It has been reported to be associated with a fetal loss rate of 1 in 770 by experienced practitioners [6]. Another diagnostic procedure, called chorionic villus sampling (CVS), involves removal of a small amount of chorionic villi from the placenta, either transcervically or transabdominally, to obtain fetal cells for testing. Although the miscarriage risk associated with CVS is greater than that of amniocentesis at most centers, the advantage is that CVS can be performed in the first trimester of pregnancy at 10–13 weeks of gestation [7]. A new screening test for Down syndrome, performed by isolating cell-free fetal DNA in the maternal circulation, may significantly reduce the number of invasive diagnostic procedures performed. Early publications indicate the sensitivity of this maternal blood test for Down syndrome is >98 % with a specificity of 99.8 %, which compares highly favorably to traditional maternal serum screening for Down syndrome with a sensitivity of 60–93 % and specificity of 1–5 % [8].

Given the increasing sophistication of ultrasonography equipment and its routine use in pregnancy, most fetal anomalies such as neural tube defects, holoprosencephaly, abdominal wall defects, or severe cardiac abnormalities are detected prenatally and rarely present unexpectedly in the

Table 3.2 Disorders recommended for routine screening in specific ethnic groups

Ethnicity	Disease	Carrier frequency	Recommended by
African, Mediterranean, Southeast Asian, Middle Eastern	Alpha thalassemia	1 in 3 Africans	ACOG
African, Mediterranean, Middle Eastern, Asian Indian	Sickle cell anemia	1 in 12 African Americans	ACOG
Northern African, Southern European, Asian Indian	Beta thalassemia	1 in 12 Southern Europeans	ACOG
Ashkenazi Jewish	Tay-Sachs	1 in 30	ACMG, ACOG
	Canavan	1 in 50	ACMG, ACOG
	Familial dysautonomia	1 in 32	ACMG, ACOG
	Bloom syndrome	1 in 100	ACMG
	Fanconi anemia (type C)	1 in 89	ACMG
	Gaucher	1 in 15	ACMG
	Niemann-Pick (type A)	1 in 90	ACMG
	Mucolipidosis IV	1 in 127	ACMG
All ethnicities	Cystic fibrosis	1 in 25 Caucasians	ACMG, ACOG
All ethnicities	Spinal muscular atrophy	1 in 35 Caucasians	ACMG

delivery room. Thus, prenatal genetic counselors often find themselves providing crisis counseling for individuals who have just been informed about a significant birth defect following a "routine ultrasound examination to confirm the pregnancy's dates." In fetal abnormality cases, when CVS or amniocentesis is performed, consideration is given to ordering comparative genomic hybridization (CGH) instead of just chromosome analysis since CGH can detect relatively small regions of deleted or duplicated genetic material compared with standard chromosome analysis [9].

Screening tests for single-gene disorders are recommended for expectant individuals or those planning a pregnancy in "high-risk ethnic groups" (see Table 3.2). The specific criteria used to select tests for population screening is based on a number of factors such as incidence in the population to be screened, medical knowledge of the disorder, and sensitivity and specificity of testing methods. In 2001, screening for cystic fibrosis (CF) was recommended but was controversial for the following three reasons: (1) the majority of expectant individuals would need to be offered screening; (2) the screening panel would not detect all carriers and would have variable detection rates depending on ethnicity; and (3) insufficient numbers of genetic counselors were available to provide informed consent [10]. Nevertheless, cystic fibrosis screening was successfully implemented by obstetricians and other primary care providers and couples who screened positive were referred to a genetic counselor.

In 2011, in dramatic divergence from ACOG and the American College of Medical Genetics (ACMG) testing guidelines, several direct to consumer marketing companies began promoting carrier screening tests for expectant couples for hundreds of exceedingly rare conditions, many of which were unfamiliar for most healthcare providers. Thus, providing proper informed consent for such prenatal panels was and remains difficult, at best. Furthermore, when a woman learns she is a carrier for a rare condition, a gene sequencing test may need to be offered to her reproductive partner since many of the mutations targeted on such panels represent a small percentage of the causative mutations for the specific condition. This may present two additional problems: (1) there may not be a clinical laboratory specifically performing sequence analysis for the rare genetic disorder in question, and (2) even if one succeeds in finding such a laboratory to perform sequencing, the reproductive partner may be found to have a variant of uncertain significance. This uncertainty may lead to confusion and high anxiety for a couple who thought that they were undergoing routine prenatal screening and would receive clear and clinically interpretable results.

Preimplantation genetic diagnosis (PGD) offers an alternative to prenatal testing for couples who are at risk of having a child with a genetic condition. Through in vitro fertilization (IVF), a single cell is removed from each embryo, usually on day 3 at the 8-cell stage, and tested for the specific genetic condition or familial mutation; then, the unaffected embryos are transferred back to the mother. PGD was first successfully performed in 1990 and is presently being offered for many monogenic disorders and chromosome abnormalities [11]. However, PGD is not yet routine as there are still many obstacles in the process related to the highly technical nature of the procedures and the difficulties in performing cytogenetic or molecular analysis on a single cell. The rate of pregnancy among patients undergoing IVF and PGD varies, but rarely exceeds about one third. Genetic counseling for couples considering PGD is imperative. Couples need to understand the risks and benefits of these

complex procedures and the likelihood of a successful pregnancy. Couples need to weigh these factors against those of standard prenatal diagnosis procedures. IVF and PGD are very expensive, approximately $15,000 per cycle, and are not covered by most insurance providers.

Prenatal genetic counselors explain the risks for various genetic conditions, present patients with prenatal diagnostic testing choices, and discuss management and outcome options. They help patients make informed and autonomous choices by encouraging exploration of personal, spiritual, and cultural beliefs that affect decision making [12, 13]. Genetic counselors support patients who choose to continue affected pregnancies by arranging appointments with pediatric specialists, fetal and pediatric surgeons, and neonatal intensive care physicians to help the family prepare for the delivery and offer to arrange contact with other families who have had a baby with the same condition. Genetic counselors also support patients who choose to terminate affected pregnancies by making the necessary referrals for the procedure, encouraging autopsy when the diagnosis is still in question, and providing referrals to support groups for individuals who have had a therapeutic abortion.

Pediatric and Adult Setting

Genetic conditions can occur with unique symptoms at all stages of life, from birth defects in a newborn to cognitive changes in an older adult. When a baby is born with birth defects or an individual at any age develops symptoms, the first step in clinical care is establishing an accurate diagnosis. Often medical geneticists are consulted by physicians caring for babies born with birth defects or other clinical symptoms to determine if there is one unifying syndrome or diagnosis that provides an explanation. Providing a diagnosis is helpful as it enables one to predict if the condition is associated with other problems that may develop over time, such as learning problems, behavioral disorders, cancers, or other medical conditions that are not present at the time of the examination. This may allow for early intervention, therapy and medical screening to reduce the impact of, or risk associated with, the conditions. Early diagnosis also may provide an explanation for likely causes of the condition and, therefore, potential recurrence risk for siblings or offspring of the affected individual. Up until 2010, chromosome analysis was a first line test in the evaluation of developmental disabilities, autism or multiple congenital anomalies. CGH should be the initial test for these conditions because the detection of causative deletions or duplications at 15–20 % is much higher than with chromosome analysis (3 %) [14].

Some individuals are affected with what appears to be a genetic condition, yet even after undergoing many tests, the diagnosis remains elusive. Exome sequencing has been conducted for such individuals on a research basis with a 25 % success rate in determining a diagnosis [15]. In 2012, exome sequencing also began to be offered on a clinical basis. Usually, in addition to sequencing the exome of the affected child, the exomes of the parents also are sequenced to determine if the child has de novo mutations or recessive mutations in the same gene from each parent. Analyzing such a large amount of data is very complex and requires an experienced bioinformatics specialist. Genetic counselors often are involved with developing the consent forms for exome sequencing as well as consenting the family members participating in exome sequencing research or clinical testing. Genetic counseling for exome sequencing is very time consuming because the genetic counselor, the patient, and their family members must consider what types of genetic variant information they are interested in learning about and are prepared to hear. For instance, in addition to the variants identified that may be causative for the disease in question, mutations predisposing to other conditions such as cancer, heart disease, and dementia may be detected.

When newborn screening for phenylketonuria was first available in the mid-1960s, the criteria developed to determine if a disorder should be considered for newborn screening included the following: (1) an acceptable treatment protocol is in place that changes the outcome for patients diagnosed early, (2) the condition's natural history is understood, (3) there is an understanding about who will require treatment, and (4) testing is reliable for both affected and unaffected patients and is acceptable to the public [16]. Many states screened for only a few conditions up until the late 1990s when tandem mass spectrometry (MSMS) technology was developed for newborn screening; then the number of conditions screened expanded dramatically. Now, most states screen for at least the 29 rare, mostly metabolic, conditions recommended by ACMG in 2005 [17]. Many of the new conditions being screened do not satisfy all of the criteria originally used for inclusion in newborn screening protocols. Genetic counselors often coordinate, or are heavily involved with, state newborn screening programs and provide valuable information to pediatricians or other healthcare providers caring for newborns who have a positive screen for one or more of these rare disorders.

The increasing access to and availability of genetic testing has improved the diagnostic capabilities for many disorders. Diagnosis of a genetic condition brings emotional, social, and financial burden for the patient and the family. Unlike many other areas of medicine, genetics has medical implications beyond the patient, extending to the entire family. Whether a diagnosis is made during the neonatal, pediatric, or adult years, the importance of genetic counseling remains. The goals of genetic counseling for the patient and

Table 3.3 Elements of pretest predictive genetic counseling [20]

Obtain family history and confirm diagnoses
Review natural history and inheritance of condition, as well as a priori risk
Discuss the benefits, limitations, and risks of testing, and the confidentiality of test results
Discuss motives for testing, anticipated result, psychosocial preparedness, and support system
Present alternatives to testing and assure that testing is voluntary and informed consent is provided

the family following the diagnosis of a genetic condition include: education about the natural history of the condition and medical implications; explanation of the genetic cause, mechanism of inheritance, and recurrence risks; identification of appropriate social and emotional resources; attentiveness to the patient's and the family's reactions to the diagnosis and their coping strategies; promotion of the best possible emotional adjustment for the patient and family; and facilitation of access to necessary medical and social services [18].

Genetic testing for diagnostic purposes is occurring more frequently without pretest counseling when ordered by a primary care provider or a specialist; however, once a positive test result is disclosed, patients are referred for genetic counseling to assist them with understanding the meaning and implications of the test result.

Predictive genetic testing can inform individuals, prior to the onset of symptoms, that they will develop or are at increased risk to develop a hereditary disorder. Experience with predictive genetic testing for adult-onset conditions such as Huntington disease and hereditary cancer syndromes has led to the development and strong endorsement of a multidisciplinary approach to predictive genetic testing that includes pretest and posttest genetic counseling protocols [19, 20]. This approach allows the patient to explore his or her motives for testing, expectations for testing, the risks and benefits of testing, and coping strategies prior to testing. Use of this recommended counseling process is especially important when no treatment or medical intervention is available for the disorder. Most predictive testing protocols require at least two pretest counseling visits (see Table 3.3) to allow the patient time to consider the benefits and risks of testing, develop a support network during the testing process, and ensure voluntary participation in testing. Experiences with Huntington disease testing have shown that patients are at risk for adverse outcomes after the disclosure of predictive genetic test results, whether the result is positive or negative [21]. Result disclosure should be done in person by a genetic counselor or healthcare professional knowledgeable about the disorder and the implications of the test result. Also, a support person for the patient should be present at the pre- and posttest counseling visits.

Cancer Setting

Cancer genetic counseling, which developed in the early 1990s, is now the second most common area of specialization for master level-trained genetic counselors. Genetic testing for inherited cancer syndromes can be useful for diagnosis and medical management for individuals with a tumor or cancer symptoms. For instance, women with breast cancer who have an inherited mutation in *BRCA1*, a tumor-suppressor gene, may be counseled by their oncologist to consider mastectomy and chemotherapy instead of lumpectomy and radiation. Furthermore, other at-risk family members could have targeted testing for the familial mutation and those who test positive may consider undergoing risk-reducing prophylactic mastectomy and oophorectomy. Chapters 22–30 highlight the various issues related to molecular testing for hereditary cancer syndromes, which include variable clinical utility, complex medical management options, dilemmas with molecular testing approaches, and testing in the research setting or during early transition of a test to the clinical laboratory. The potential risks and benefits associated with testing vary based on the specific hereditary cancer syndrome, as well as the patient and family history. The American Society of Clinical Oncology recommends pre- and posttest counseling for individuals referred for cancer genetic testing [22]. The genetic counselor discusses the details of the genetic testing (detection rate, clinical utility, recurrence risk, etc.), as well as early detection and prevention options for individuals with a positive test result. Often, hereditary cancer syndromes increase an individual's risk for cancer in multiple organ systems, which makes medical management, screening, and early detection more complex. For example, some individuals at risk for von Hippel-Lindau syndrome need at least annual screening for brain and spine hemangioblastomas, retinal angiomas, pheochromocytoma, renal cell carcinoma, and other tumors.

Informed Consent

In the healthcare setting, the process of informed consent is a protection for patients. Prior to diagnostic testing or therapeutic intervention, the provider explains the procedure to the patient, along with the risks, benefits, and alternatives, so that the patient can voluntarily make informed decisions about diagnostic and treatment options [23]. Depending on state law and laboratory standards, variation in informed consent requirements for genetic testing exist. Position statements and guidelines for informed consent for genetic testing are available for a growing number of conditions or groups of conditions. The majority of guidelines address issues related to predictive genetic testing. The NSGC recommends

Table 3.4 Key elements of informed consent for genetic testing

Discussion of purpose of the test and risks of procedures involved in obtaining a sample for testing
Clinical utility of the test and interpretation of all possible test results (positive, negative, uncertain, test failure)
Discussion of risks, benefits, and limitations of testing (including psychosocial, cultural, and financial)
Presentation of alternatives to genetic testing
Description of the procedure for communication of results
Confidentiality of test results
Voluntary nature of informed consent

obtaining informed consent prior to predictive genetic testing for adult-onset conditions. Guidelines for informed consent prior to genetic testing stress that this is more than having a patient read and sign a piece of paper. Informed consent should be a communication process that fosters autonomous and informed decision making by the patient [24, 25]. Presentation of the key elements of informed consent (Table 3.4) needs to be tailored to the individual patient's learning style, educational and cultural background, and family situation to optimize the usefulness of informed consent for the patient and their family. This is a time-intensive process that cannot be done by primary care physicians during a routine office visit. The informed consent process also applies to collection of tissue or body fluids for genetic or other research purposes [26].

Genetic Testing for Children and Adolescents

The benefits and harms of genetic testing need to be carefully evaluated before proceeding with testing in children who may not be able to appreciate the implications of such results. When genetic testing directly impacts medical management or treatment for a child with symptoms or clinical features of a condition, the benefits of testing are clear and the well-being of the child is being promoted. However, when genetic testing does not impact medical management, or the condition in question will occur in adulthood, the implications of testing become more complex and the benefits become less clear. The ACMG and the American Society of Human Genetics wrote "Points to Consider: Ethical, Legal and Psychosocial Implications of Genetic Testing in Children and Adolescents" [27]. The recommendations are the following:

1. Timely medical benefit to the child should be the primary justification for genetic testing.
2. Substantial psychological benefits to the competent adolescent also may be a justification for genetic testing.
3. If the medical or psychological benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset diseases, genetic testing should be deferred.
4. If the balance of benefits and harms is uncertain, the provider should respect the decision of the competent adolescent and his or her family.
5. Testing should be discouraged when the provider determines that the potential harms of genetic testing in children and adolescents outweigh the potential benefits.

Education and counseling for the parents and the child, at an appropriate level, should be provided. The benefits and harms related to medical issues, psychosocial issues, and reproductive issues need to be presented and discussed. Children and certainly adolescents have decision-making capacity. The child's competence and wishes should be assessed prior to genetic testing and carefully balanced with parental authority. This is especially true for adolescents who can articulate a specific opinion that differs from that of his or her parents [27]. Assent from the child or adolescent should be obtained in addition to informed consent from the parents.

Legal Protection

In the past decade, there was great concern about the risk for health insurance discrimination related to testing for predisposition to various familial cancer syndromes and adult onset conditions such as Huntington disease. With the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008, a federal law that protects Americans from being treated unfairly because of differences in their DNA that may affect their health, this concern has largely been put to rest as GINA provides protection against health insurance and employment discrimination. Even so, GINA does not provide protection against life insurance, disability insurance, or long-term care insurance discrimination [28]. Furthermore, GINA does not provide protection for federal employees or those who are employed by companies with less than 15 individuals. Genetic counselors need to explore whether their patients are protected from health and employment discrimination and incorporate this into the discussion of risks and benefits of genetic testing.

Other Roles for Genetic Counselors

Research Testing

Genetic counseling can be valuable to individuals who are participating in research genetic testing. Research participants may have difficulty understanding the purpose, risks,

and benefits of the study. In addition, many participants have expectations of receiving research testing results and do not appreciate the limitations of reporting results in the research setting. Therefore, consent forms created by genetic counselors in language understandable to the general public are a critical component of genetic research. Disclosure of genetic research results to participants is optimally done by a professional able to provide genetic counseling. For many rare conditions, clinical testing is not available; therefore, research testing is the only option for families. When genetic testing is transitioning from research to the clinical setting, a genetic counselor can be a liaison between the participant and the laboratory and be responsible for informed consent and disclosure of results.

Genetic Counselors Working in the Laboratory

Genetic tests differ in many ways from other laboratory tests. One difference is the necessity for the laboratory to receive clinical information, including the patient's symptoms and family history, for proper interpretation of test results. Genetic counselors understand the clinical information needed by molecular laboratories to interpret specific genetic tests. Additionally, genetic counselors can review the clinical information provided to determine whether the test ordered is the most appropriate and cost-effective test given the patient's symptoms and family history. One study of genetic counselors who reviewed thousands of genetic test orders for a laboratory over a 10-month period revealed that greater than one-third of complex genetic tests were misordered [29]. Oftentimes, other healthcare providers do not understand the importance of obtaining medical records documenting a familial condition. For example, if a patient requests testing for spinocerebellar ataxia (SCA) because this condition has been diagnosed in a sibling, the specific type of SCA must be determined, as there are currently 17 causative genes and commercial testing is available for only about 9 of the 17 genes. If the individual requesting the test does not know the specific SCA type, one could order testing for all of the commercially available types; however, a negative result would not eliminate the individual's risk because the proband may have one of the SCAs not included in the testing. A genetic counselor can work with an at-risk individual to obtain the necessary family history information and documentation so that the most accurate and efficient approach to testing can be used and the interpretation of the results will be more informative. An accurate and comprehensive family history is a valuable tool in a diagnostic evaluation as it can be used as a medical screening tool, establish a pattern of inheritance, identify individuals at risk, and determine strategies for genetic testing.

Understanding the clinical validity of a genetic test result can be difficult for both healthcare providers and patients. However, the sensitivity and specificity of the test method used by the testing laboratory are paramount to the interpretation of the results [12]. Many molecular, biochemical, and cytogenetic laboratories have genetic counselors on staff who can be a useful resource for other healthcare providers and the public. They are able to provide the necessary education and information to determine the appropriate approach to genetic testing, facilitate the details of ordering a test, and help interpret and communicate test results (Table 3.5).

Table 3.5 Genetic counselors working in the laboratory

Obtain test-specific clinical information and family history to ensure that the most appropriate test is performed
Facilitate or document the informed consent process to assure that the patient's autonomy is protected
Assist referring physicians and patients with understanding the test results and implications for the patient and family members
Facilitate collection of appropriate family samples for clarifying the significance of variants of uncertain significance

Summary

The goals of genetic counseling are to address the informational and emotional needs of patients and their families [13]. For example, the explanation of risks and benefits associated with a genetic test that is tailored to a patient's educational needs as well as their family, social, and cultural background facilitates informed decision making by promoting patient autonomy and informed consent. The key goals of genetic counseling for most patient encounters include the following:

- Obtain and interpret family medical history information.
- Educate patients so that they understand the medical and genetic information (inheritance and recurrence risks) needed to make health-management decisions and understand their condition.
- Promote informed decision making and informed consent.
- Be aware of nontechnical factors (social, cultural, financial, and emotional factors) that influence patients in the decision-making process.
- Foster genetic competence in patients and families.
- Identify social and professional resources for patients.

As genetic testing expands and is incorporated into mainstream healthcare, especially for disease prevention and treatment (pharmacogenetics) for common disorders, pre- and posttest genetic counseling for every test will not be feasible. However, genetic counseling services need to be available to assist healthcare providers and patients with education and support facilitating safety in genetic testing.

References

1. Sarah Lawrence College Graduate Programs; History. http://www.slc.edu/graduate/programs/human-genetics/character/history/index.html. Accessed 20 Jan 2012.
2. Genetic counseling Programs-US; Accreditation of Training; Programs; http://www.abgc.net/Training_Program_Accreditation/US_Accredited_Programs.asp. Accessed 18 Jan 2012.
3. 2010 Professional Status Survey: Executive Summary; Primary Specialty Area. http://www.nsgc.org/Portals/0/Publications/PSS%202010%20Executive%20Summary%20FINAL.pdf. Accessed 18 Jan 2012.
4. Resta R, Biesecker BB, Bennett RL, et al. A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. J Genet Couns. 2006;15(2):77–83.
5. ACOG Practice Bulletin. Clinical Management Guidelines for Obstetrician—Gynecologists. Number 77, Jan 2007.
6. Odibo AO et al. Revisiting the fetal loss rate after second-trimester genetic amniocentesis: a single center's 16 year experience. Obstet Gynecol. 2008;111:589–95.
7. Shulman LP, Elias S. Amniocentesis and chorionic villus sampling in fetal medicine. West J Med. 1993;159(special issue): 260–8.
8. Palomaki GE, Kloza EM, Lambert-Messerlian GM, et al. DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. Genet Med. 2011;13(11): 913–20.
9. Fiorentino F, Caiazzo F, Napolitano S, et al. Introducing array comparative genomic hybridization into routine prenatal diagnosis practice: a prospective study on over 1000 consecutive clinical cases. Prenat Diagn. 2011;31(13):1270–82.
10. National Institutes of Health Consensus Development Conference Statement on genetic testing for cystic fibrosis. Arch Intern Med. 1999;159:1529–39.
11. Kanavakis E, Traeger-Synodinos J. Preimplantation genetic diagnosis in clinical practice. J Med Genet. 2002;39:6–11.
12. Burke W. Genomic medicine: genetic testing. N Engl J Med. 2002;347:1867–75.
13. Weil J. Genetic counseling in the era of genomic medicine: as we move towards personalized medicine, it becomes more important to help patients understand genetic tests and make complex decisions about their health. EMBO Rep. 2002;3:590–3.
14. Miller DT, Adam MP, Swaroop A, et al. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010;86:749–64.
15. Gahl WA, Markello TC, Toro C, et al. The National Institutes of Health Undiagnosed Diseases Program: insights into rare diseases. Genet Med. 2012;14(1):51–9.
16. Ross LF. Screening for conditions that do not meet the Wilson and Jungner criteria: the case of Duchenne muscular dystrophy. Am J Med Genet. 2006;140A(8):914–22.
17. Natowicz M. Newborn screening—setting evidence-based policy for protection. N Engl J Med. 2005;353:867–70.
18. Walker AP. The practice of genetic counseling. In: Baker DL, Schulette JL, Uhlmann WR, editors. A guide to genetic counseling. New York: Wiley-Liss; 1998. p. 1–20.
19. Almqvist EW, Bloch M, Brinkman R, Craufurd D, Hayden MR. A worldwide assessment of the frequency of suicide, suicide attempts, or psychiatric hospitalization after predictive testing for Huntington disease. Am J Hum Genet. 1999;64:1289–92.
20. International Huntington Association and World Federation of Neurology. Guidelines for the molecular genetic predictive test in Huntington's disease. J Med Genet. 1994;31:555–9.
21. McKinnon WC, Baty BJ, Bennett RL, et al. Predisposition genetic testing for late-onset disorders in adults: a position paper of the National Society of Genetic Counselors. JAMA. 1997;278:1217–20.
22. American Society of Clinical Oncology (ASCO). Policy statement update: genetic testing for cancer susceptibility. J Clin Oncol. 2003; 21:2397–406.
23. Andrews LB. Compromised consent: deficiencies in the consent process for genetic testing. JAMA. 1997;52:39–44.
24. Geller G, Botkin JR, Green MJ, et al. Genetic testing for susceptibility to adult-onset cancer: the process and content of informed consent. JAMA. 1997;277:1467–74.
25. Sharpe NF. Informed consent and Huntington disease: a model for communication. Am J Med Genet. 1994;50:239–46.
26. American Society of Human Genetics (ASHG) Report. Statement on informed consent for genetic research. Am J Hum Genet. 1996;59:471–4.
27. American College of Medical Genetics/ASHG. Points to consider: ethical, legal and psychosocial implications of genetic testing in children and adolescents. Am J Hum Genet. 1995;57:1233–41.
28. Clifton JM, VanBeuge SS, Mladenka C, et al. The Genetic Information Nondiscrimination Act 2008: what clinicians should understand. J Am Acad Nurse Pract. 2010;22(5):246–9.
29. Miller C. Value of genetic counselors in the laboratory. Adv Med Lab Prof http://laboratorian.advanceweb.com/features/articles/value-of-genetic-counselors-in-the-laboratory.aspx?CP=2.