Genetic Counseling in Pediatrics

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Education Gap

Physicians should be aware that genetic counselors can serve as a trusted resource for both physicians themselves as well as their patients and families. Genetic counselors are skilled in the areas of health-care communication, patient education, risk analysis, the genetic testing process, and family support.

Objectives

After completing this article, readers should be able to:

1. Discover the role of a genetic counselor in a pediatric setting.
2. Recognize circumstances in which genetic testing may be beneficial.
3. Differentiate the roles and training backgrounds of medical geneticists and genetic counselors.
4. Understand when to refer a patient to a geneticist or genetic counselor.

Abstract

Genetic counseling is a communication process whereby an individual or family obtains information about a genetic condition, is helped to understand the implications and significance of the condition, and is given resources to help with coping and management. It is a continuous process involving lasting supportive relationships between the family and the genetic professional. Genetic counselors are master's level–trained health-care professionals who work closely with pediatricians and pediatric subspecialists alike. Genetic counselors can be a source of information about genetic conditions, risk assessment for disease, and genetic testing. Although most of a genetic counselor's job is patient care and education, genetic counselors also serve as resources to educate health professionals about genetics.

AUTHOR DISCLOSURE Mr Stein and Mss Loman and Zuck have disclosed no financial relationships relevant to this article. This commentary does not contain a discussion of an unapproved/investigative use of a commercial product/device.

INTRODUCTION

A 3-year-old boy was born with a cleft palate that was successfully repaired at 1 year of age. He has been generally healthy since but has yet to develop expressive speech. His parents and older siblings are in good health with normal intellect. Could the cleft palate and lack of expressive speech be related? How could one find out? Is there a genetic test that should be performed? What is the impact to family members if a genetic diagnosis is made? How could genetic counseling benefit the family?

In cases such as this, a genetics consultation, involving the team of a geneticist and a genetic counselor, is an appropriate next step given the cleft palate and lack of speech. Together, with the family, they can decide whether genetic testing is medically indicated and if so, which type. If genetic testing is pursued and the results are informative for making a diagnosis, tailored genetic counseling, including education, resources, and psychosocial support, can be provided to the family.

Genetic counselors are a part of health-care teams and provide information to patients in a variety of different settings and specialties. Pediatrics is one of the most common areas of clinical practice for a genetic counselor and is a focus of this article.

GENETICS, GENETIC COUNSELING, AND PEDIATRIC GENETIC COUNSELORS

Genetic counselors are integrated into the pediatric setting by providing direct patient care (ie, meeting face-to-face with families) and by serving as a reliable resource to pediatricians, nurses, administrators, and other members of the health-care team. Historically and currently, the most common setting for the pediatric genetic counselor has been working alongside a medical geneticist in a medical genetics clinic. This setting, however, is evolving and some genetic counselors now work as closely with other pediatric providers, such as neonatologists, neurologists, pulmonologists, cardiologists, oncologists, or developmental pediatricians—all of whom see patients with genetic conditions in their practice and/or are involved in ordering genetic testing (Table 1). No matter the setting or the scenario, a variety of core components are typically included in the genetic counseling process, including family and medical history interpretation, patient education, psychosocial assessment, and facilitated decision making.

Medical and Family Histories

One core aspect of genetic counseling, *information gathering*, is most commonly performed as the collection of family and medical histories. Genetic counselors typically obtain detailed family histories that involve eliciting and creating a 3-generation pedigree, which is subsequently added to the medical record. The eliciting of the pedigree includes obtaining general health information on family members as well as asking targeted questions specific to the referral indication. For example, if a patient were to be referred for an indication of multiple café-au-lait macules with concern for neurofibromatosis type 1 (NF1), then the eliciting of the family history should include targeted questions about other family members' potential features of NF1 as well as similar genetic conditions. In this situation, a genetic counselor will ask whether family members have café-au-lait macules or "birthmarks" similar to the patient; if there is a family

TABLE 1. **Examples of Genetic Conditions, by Pediatric Subspecialty, that Warrant the Involvement of a Genetic Counselor**

PEDIATRIC SUBSPECIALTY	SAMPLE GENETIC CONDITIONS WARRANTING GENETIC COUNSELING
Cardiology	Long QT syndrome Marfan syndrome
Dermatology	Ichthyosis Tuberous sclerosis
Developmental-behavioral	Fragile X syndrome Rett syndrome
Endocrinology	SHOX deficiency disorders Turner syndrome
Gastroenterology	Alagille syndrome Turner syndrome
Hematology	Sickle cell disease Hemophilia
Immunology	Chronic granulomatous Severe combined immunodeficiency
Nephrology	Alport syndrome Nephrogenic diabetes insipidus
Neurology	Friedreich ataxia Muscular dystrophy
Oncology	Li-Fraumeni syndrome von Hippel-Lindau syndrome
Ophthalmology	Leber congenital amaurosis Retinitis pigmentosa
Otolaryngology	22q11.2 deletion syndrome Usher syndrome
Pulmonology	Cystic fibrosis Primary ciliary dyskinesia
Rheumatology	Ankylosing spondylitis Ehlers-Danlos syndrome

history of any tumors, "lumps or bumps" of any sort; if there is any freckling in the groin or arm pits of any family members; or if learning disabilities are present in the family because these can all be characteristics associated with NF1. The genetic counselor will also ask about family history of hearing loss or ringing in the ears because these can be features of NF2, a similar condition. Once a detailed family history is obtained, it is assessed, analyzed, and interpreted to aid in determination of the likelihood of a genetic condition in the patient, the most likely inheritance pattern, and the chances of recurrence for family members. Medical history is interpreted in the context of family history to help assess the likelihood of a genetic diagnosis. For those with an established diagnosis, family history information is used to help determine chances of recurrence. This includes risk calculation, risk assessment, and sometimes Bayesian analysis to determine the chances of family members or any future offspring being affected with the same condition.

Family history and medical information are also used to help determine the necessity of genetic testing for both the patient and family members. If other family members have previously had genetic testing for a specified condition, this information is essential in determining the appropriate testing for a patient for that condition. For example, with NF1, if the proband (ie, the person serving as the starting point for the genetic study) has had full sequencing of the *NF1* gene, then the pedigree can be used to determine which family members may benefit from targeted variant testing and for whom genetic testing would be contraindicated based on inheritance. This is significant given that there is great variability and expense associated with genetic testing.

Patient Education

Individuals or families with a genetic condition (or at risk for a genetic condition) commonly seek information about the diagnosis itself, prognosis, management, and implications to the family, thus making education an essential and multifaceted element of genetic counseling. It involves talking with families about inheritance patterns, helping families to understand how this information may impact them, and what it all means for their family. Patient education also involves teaching families about genetic conditions, their features, long-term prognosis, and management/treatment options. Even before a diagnosis, families are educated about any genetic testing that may be recommended for the patient. This includes a variety of aspects such as the name of the test, what it will test for, what it will not test for, limitations of the test, and the possible results of the test.

In the genetic counseling setting, the genetic counselor will often have a preplanned outline of topics considered essential to cover. Depending on the referral indication, this outline may include the condition name, symptoms, prognosis, diagnostic tools, formal genetics, genetic testing, recurrence risks, and/or inheritance patterns. For any educational topic, visual aids are crucial in a genetic counseling session to help orient people's attention as well as helping with patient understanding. This is especially true with numbers and probabilistic information. (1) Pictures, charts, and other visuals are a way to help translate complex genetic topics into something patients and their families can better understand. Similarly, genetic counselors will often also try to use analogies to help families better understand genetic concepts. When discussing inheritance patterns, the pedigree itself can be used as a visual aid.

At its core, genetic counseling is built on the essential tenet of effective communication, that is, communicating complex information with clarity in both the oral and written forms. To that end, genetic counselors regularly provide families with written information and resources. Because genetic topics can be immensely complex, summary letters or "patient letters" are sometimes sent to families after an appointment with a genetic counselor. These letters provide families with a summary of the pertinent information that was discussed at the visit and can serve as a resource to be referred back to later or shared with other family members. (2)

Genetic counseling itself is not necessarily confined to a single visit. At an initial appointment, a genetic counselor may only briefly discuss the suspected condition but then go into more detail once an official diagnosis is established. The initial shock of a new diagnosis may make it difficult for a family to fully comprehend the information in a single visit. Furthermore, the very nature of the profession also means that sometimes genetic counselors must deliver upsetting or bad news. This news may sometimes be better absorbed in multiple steps.

For some individuals, genetic counseling occurs across the lifespan. A prime example is individuals/families with metabolic conditions diagnosed by newborn screening. Initially, the educational components focus on the parents and their needs. Over time, the child may become a more active participant in his or her own health-care and wonder why he or she has to attend medical visits or eat foods different from their friends. Pediatric genetic counselors are trained in translating difficult genetic concepts into explanations that parents and children of varying educational levels and different developmental stages can understand.

Psychosocial

Helping a family cope with a genetic diagnosis or with the continuing uncertainty of searching for one can be

challenging. Because of this, an understanding of the psychosocial consequences of a genetic diagnosis is important to provide appropriate assistance. Fear and worry are common feelings of parents of children with genetic diagnoses, as are guilt, shame, and uncertainty. Genetic counselors are trained to work with families who may be feeling this way. Listening and support are crucial to understand how a patient or a family member is feeling and to better understand the impact that a genetic condition is having on the patient/family. Genetic counselors provide families with a space to share their experiences and the impact that a genetic diagnosis, or lack of, has had on their life and work. They also help connect families with support groups, similarly affected families, and other community or national resources. In addition, they advocate on behalf of the patient in arenas such as the health and educational systems.

Patient Decision Making and Informed Consent

It is worth noting that when it comes to making decisions regarding genetic testing, family planning, or medical procedures, a genetic counselor does not direct or influence the patient on what decision to make but rather presents information in a nonjudgmental fashion—not with the purpose of encouraging or discouraging a particular course or action. In the era of precision medicine, precision consent is also necessary. In other words, genetic counselors are trained (and strive) to help patients/families make decisions that are right for them. In the pediatric setting, this often involves helping families decide whether a certain genetic test is something they would like to pursue. There are many factors that a family can consider when deciding if they want to undergo genetic testing, including the possible results of the test, how the results could change management, what undesirable information they could learn from the test, the impact of a positive or a negative result, whether having a confirmed diagnosis is important to them, how much the test costs, and whether the test will be covered by insurance. Patients/families must consent to genetic testing before it begins, and this consent should be voluntary and rooted in patient understanding of the many variables.

Genesurance Counseling

A more recent addition to the roles and responsibilities of genetic counselors centers around genesurance counseling. Genesurance counseling is that portion of a genetic counseling session, whether intentional or unintentional, that is devoted to the topic of costs and insurance/third-party coverage (particularly for genetic testing). (3) Genesurance counseling helps to educate families about the financial aspects of a genetic test, which is often an important factor for families when deciding whether they would like to pursue genetic testing. Through genesurance counseling, genetic counselors help families understand the financial aspects of any recommended genetic testing, including medical necessity, deductibles, list price, and pricing differences among laboratories. In many cases, prior authorization for a genetic test may be needed, or there may be additional steps required to determine whether a genetic test will be a covered service. Genetic counselors have found themselves helping to navigate this process for patients and their families in recent years. In addition, many families have questions surrounding release of genetic test information to third-party payers, insurability after a genetic diagnosis, potential genetic discrimination, and confidentiality. It is important for genetic counselors to stay current on the ethical, legal, and social implications of genetic testing so that they may provide the most relevant and timely information in this regard.

THE COMPLEXITIES AND NUANCES OF GENETIC TESTING

Knowing when to order a genetic test and choosing the right genetic test takes an in-depth knowledge of the nuances, functionality, and capabilities of the test. Similarly, it is necessary to know under which circumstances genetic testing may or may not be indicated.

There are several reasons why genetic testing is beneficial in the right circumstance. A primary reason is that making or confirming a diagnosis can lead directly to changes in medical management. For example, depending on the genetic condition this could mean additional hearing, vision, musculoskeletal, or heart screening. Or, it could mean eligibility for a clinical trial, treatment, or therapy. In addition, making a genetic diagnosis often helps with prognosis. Although each patient is uniquely different despite his or her diagnosis, typically a positive/informative genetic test gives some insight about what to expect for a child's future.

In some cases, genetic testing allows families to end a diagnostic odyssey, thereby giving a name to their condition previously only broadly describable by a collection of symptoms. Although some may argue that ending a diagnostic odyssey is not an extrinsically purposeful reason to pursue genetic testing because it does not change medical management, it does ultimately end up saving the family's time and financial resources. (4) In addition, families who ultimately receive a diagnosis for their child often use this information to connect with other families. Although genetic diseases are collectively common, they are

individually rare, meaning that there may be no other families in the region similarly affected. Social media, however, has made it possible for families from across the country or around the world to connect and share advice and resources, and in some cases, arrange meetings in a central location.

Advances in the robustness of genetic testing, such as exome sequencing in which thousands of genes are screened simultaneously, have made ending the diagnostic odyssey more commonplace in recent years. This type of genetic testing is expensive, however, costing thousands of dollars (Table 2). Although exome testing is appropriate in some limited circumstances, in other cases a different type of testing, such as a single-gene test, a panel test involving multiple genes, carrier testing, or site-specific testing for a known familial mutation may be the best option. Although describing the differences in these test options is beyond the scope of this article, it is important to know that each test carries with it extreme differences in pricing, robustness, accuracy, and sensitivity. Genetic counselors (and medical geneticists) are trained in determining the most appropriate approaches to genetic testing as well as in the interpretation of the positive, negative, or uncertain results that follow.

A confirmed or new diagnosis, based on genetic testing, will allow recurrence risk to be determined and may impact the family in several ways. If the condition is a sporadic event caused by a new spontaneous gene mutation (de novo mutation), the recurrence risk is low to the family. On the other hand, if the condition is deemed to be autosomal recessive, the parents then have a 1 in 4 risk with any given pregnancy to have an additional affected child and, depending on the seriousness of the condition, may want to weigh several reproductive options. These options could include deciding not to have any more children, using a donor egg, donor sperm, adoption, prenatal diagnosis, or preimplantation genetic diagnosis, which allows couples to have their own biological children and avoid prenatal diagnosis by testing embryos created by in vitro fertilization before implantation.

TABLE 2. **Relative Costs of Commonly Ordered Genetic Tests**

TYPE OF GENETIC TEST	RELATIVE COST
Targeted testing for a known familial variant	$
FISH for single chromosome location	$
Chromosome analysis/ karyotype	$$
Chromosomal microarray	$$$
Single-gene testing	$$–$$$$$$
Genetic testing panel of multiple genes	$$$–$$$$$$$$$$$$
Exome sequencing	$$$$$$$$$$–$$$$$$$$$$$$ $$$$$$$$

The cost of genetic testing varies greatly between different performing laboratories, but, in general, targeted testing for a single known familial variant is on the least expensive end of the spectrum, with exome sequencing on the more expensive end. For illustrative purposes only, each "$" in the table could be thought of as representing approximately $500 (based on market analysis and expert opinion). FISH=fluorescence in situ hybridization.

Despite compelling reasons to consider genetic testing in appropriate circumstances, there are equally as compelling reasons to avoid genetic testing for some patients and conditions, including conditions with no symptoms until adulthood or no pediatric preventive care options. One such situation is when a condition will not display symptoms until adulthood or there is no cure or screening recommendations in childhood. Examples of this include the *BRCA1* and *BRCA2* genes for hereditary breast ovarian cancer syndrome, and the *HD* gene that leads to Huntington disease. In these circumstances, the testing of minors should, in general, not be pursued. Rather, the decision to undergo genetic testing should rest with the individuals themselves once they reach legal age and ability to consent to such testing. (5) Similarly, there is generally not a compelling reason to do carrier testing of autosomal recessive disorders for minors given that carriers are asymptomatic.

Genetic counselors and medical geneticists are experts in navigating genetic testing options, interpreting results, searching the literature, and communicating the meaning of results to the physician, patient, and/or family member. In addition, they are available as a resource to health-care providers who have questions about how to approach genetic testing. These individuals not only provide assistance and resources but may also help educate health-care providers about the tools available. Furthermore, a new field of genetic counselors that has recently emerged is that of laboratory genetic counselors. These genetic counselors regularly facilitate communication between health-care professionals and the laboratory to ensure that genetic testing is used wisely and correctly (ie, utilization management) and can also assist health-care professionals in understanding complex testing results or in determining appropriate testing options for a patient. (6)

THE DIFFERENCES IN TRAINING AND ROLES BETWEEN GENETIC COUNSELORS AND MEDICAL GENETICISTS

Although geneticists and genetic counselors commonly work as a team in the pediatric setting, they come from different training backgrounds, resulting in different roles, responsibilities, and scopes of practice. Medical geneticists are physicians who have chosen genetics and genomics as a specialty. There are multiple training program routes that can be undertaken: a primary residency program in genetics (categorical residency programs), multiple residency experiences, or combined residency programs. (7) In the categorical residency, an individual must first complete at least a 1-year residency in a program that is accredited by the Accreditation Council for Graduate Medical Education. The first year can be in any specialty, but the more common examples include pediatrics, internal medicine, family medicine, or obstetrics and gynecology. After completion of the 1-year residency, an individual must then complete a 2-year medical genetics residency program with training in disciplines such as reproductive genetics, pediatric and adult genetic disorders, dysmorphology, inborn errors of metabolism, and cancer genetics. Those individuals are then eligible to take board certification examinations for genetics and genomics.

The second option commonly pursued to become a medical geneticist is to first fully complete a residency in a discipline such as a pediatrics or internal medicine then continue on to complete a second 2-year categorical residency in genetics. Individuals opting for this route are able to be board certified in both residency specialty areas.

The third option, combined genetics residencies programs, typically lasts 4 to 5 years. These residencies combine medical genetics with the fields of internal medicine, pediatrics, maternal-fetal medicine, and reproductive endocrinology and infertility. There is some variation in when a physician begins the residency program; internal medicine and pediatric programs start after the completion of medical school, and maternal-fetal medicine and reproductive endocrinology and infertility programs do not start until the completion of an obstetrics and gynecology residency.

Genetic counselors receive a master's degree from an accredited genetic counseling training program. The most common undergraduate degrees for those entering the field are in biology, genetics, and psychology, but there is not a required undergraduate major. Graduate programs in genetic counseling consist of a combination of classroom training and clinical rotations in multiple specialty areas. The classroom training focuses on courses in medical genetics, laboratory genetics, communication, and counseling. Programs often have their own unique, qualities such as curriculum in bioethics, cultural competency, or community engagement. Field practicum experiences vary between programs, but the 3 core specialties consist of prenatal, cancer, and pediatric experiences. As the field of genetic counseling has continued to develop into new specialties, clinical rotations have also emerged in other specialties, such adult genetics, cardiology, neurology, laboratory counseling, and even primary care. In addition to coursework and clinical rotations, genetic counseling graduate students must complete a graduate research project. Most often, these projects are original research projects that can be used to further the field of medical genetics by responding to current questions and needs of the field.

Differences in training and scope of practice allow medical geneticists and genetic counselors in the pediatric setting to have complementary roles. Medical geneticists' roles during an appointment include performing the clinical examination, providing recommendations for testing, ordering and managing medications, and interpreting biochemical results and imaging. Genetic counselors are then able to focus more on family and medical history, education of genetic information, and psychosocial concerns but leave all physical examinations to the medical geneticist.

When to Refer to Medical Genetics

Understanding the differences between geneticists and genetic counselors is necessary in determining the appropriate referral. In the pediatric setting, it is uncommon for a patient to meet solely with a genetic counselor with no involvement by a medical geneticist or other provider. This is because many of these children require an examination as part of the complete evaluation. Most medical genetics practices have genetic counselors working as part of their team. Following are some examples of common reasons for referral to a medical geneticist.

Example 1. *In the clinic today, you saw a 4-year-old boy for a health supervision visit with 7 café-au-lait macules. When you discussed this finding with his mom, she stated that she did not think anything of them because she and her mom have the same "birthmarks." As a pediatrician, you know that this is a good patient to refer due to his physical findings and the possible family history of NF1. Clinical examination and discussion of genetic testing options would be beneficial to the family to determine the risk of additional features related to NF and future management recommendations. A referral to a medical genetics clinic is warranted.*

Example 2. *You are seeing an 8-month-old girl who was born with a congenital heart defect, hypotonia, failure to thrive due to feeding difficulties, and has been delayed in meeting her motor milestones. She recently started rolling 1 direction but is not able to sit without support. Referral to genetics for patients such as this can be helpful in determining whether there is a single*

underlying cause. It is likely that an assortment of tests would be considered or needed to determine whether this child's concerns are related.

When to Refer to a Genetic Counselor

In some situations, a referral directly to a pediatric genetic counselor can be useful. This is especially true if the family previously received a diagnosis but requires more time to discuss the results and condition. Another example includes patients with a family history of a genetic syndrome or a previously affected child who are looking for carrier screening or further discussion of reproductive risks.

When a medical provider is in doubt regarding whether to refer a patient for a medical genetics visit or genetic counseling only visit, it is always appropriate to call and speak to a genetic counselor. By providing the genetic counselor with basic information, he or she is able to help the provider determine the appropriate referral type. For those not familiar with the genetics services in their area, including genetic counselors, a searchable and useful resource is the "Find a Genetic Counselor" tool on the National Society of Genetic Counselors website (searchable by location and specialty type). (8)

Summary

- Genetic counselors are integrated into the pediatric setting by providing direct patient care (ie, meeting face-to-face with families) and by serving as a reliable resource to pediatricians, nurses, administrators, and other members of the health-care team.
- Individuals or families with a genetic condition (or at risk for a genetic condition) commonly seek information about the diagnosis itself, prognosis, management, and implications to the family, thus making education an essential and multifaceted element of genetic counseling.
- Genetic counseling is not the same as genetic testing but rather is a process by which complex genetic information is communicated to patients in an easily understood way.
- At its core, genetic counseling is built on the essential tenet of effective communication—both orally and in writing.
- Informed consent for genetic testing should be voluntary and rooted in patient understanding.
- A genetic counselor presents factual information in a balanced and understandable manner in order for the patient and family to make informed decisions about testing and management, not with the purpose of encouraging or discouraging a particular outcome or decision.

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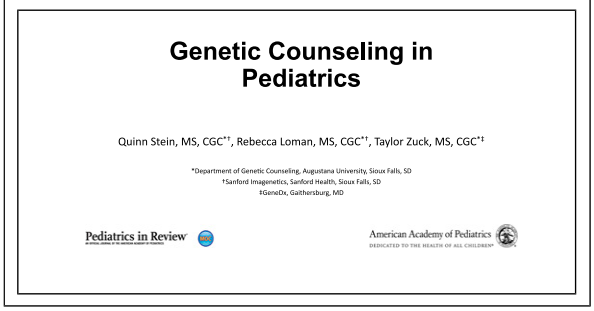

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1. A child with multiple café-au-lait spots is referred to the genetics clinic by his primary care physician. The genetic counselor begins the visit with information gathering. Which of the following is the most appropriate part of the information gathering that is conducted by the genetic counselor on this patient?
    A. A 3-generation pedigree of targeted family and medical histories.
    B. Family history of cleft lip with or without cleft palate.
    C. History of environmental exposures of patient and family members.
    D. Magnetic resonance imaging of the brain of the patient and his first-degree relatives.
    E. Punch biopsy of the skin lesions.
2. A 3-year-old boy with infantile spasms, shagreen patches, confetti lesions, and cortical tubers on magnetic resonance imaging is seen in the clinic and diagnosed as having tuberous sclerosis. The managing physician refers him to a genetic counselor for follow-up. Which of the following is the most appropriate area in which the genetic counselor will play a major role in the care of this patient?
    A. Assist the family with paternity testing.
    B. Educate the parents by reviewing the patient's clinical diagnosis, the natural history, and the inheritance pattern of tuberous sclerosis.
    C. Manage the patient's infantile spasms.
    D. Order genetic testing for tuberous sclerosis for the patient's developmentally normal and clinically asymptomatic 1-year-old female sibling.
    E. Wood lamp examination of both parents looking for skin findings of tuberous sclerosis in either parent.
3. A newborn female is diagnosed as having phenylketonuria on newborn screening. Confirmatory testing is ordered and the baby is started on a special formula low in phenyalanine. The managing physician discusses the diagnosis with the family and refers the baby to the genetic and metabolic clinic for continued follow-up after discharge from the hospital. At the clinic, the patient and family are seen by a geneticist and a genetic counselor. The genetic counselor is expected to monitor this patient until which of the following times in his life?
    A. Across the lifespan of the patient.
    B. One year of age to wean off the special formula.
    C. Six months of age to educate the family about starting solid foods.
    D. Until the patient starts school.
    E. Until the start of the first menstrual period.
4. An 8-year-old boy is seen in the clinic with lens dislocation, aortic root enlargement, and tall stature with long arms. He is diagnosed as having Marfan syndrome. The family is stressed out about the new diagnosis and seems to have many questions. You explain to the parents that the patient will be referred to multiple specialists who will be following him. Which of the following health professionals is more likely to connect the family with support groups related to the new overall diagnosis?
    A. Cardiologist.
    B. Child life specialist.
    C. Genetic counselor.
    D. Ophthalmologist.
    E. Psychiatrist.

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5. You are following a 5-year-old girl with short stature and pulmonic stenosis. You suspect Noonan syndrome and refer the family to medical genetics for consultation. The family meets with a clinical geneticist and a genetic counselor. DNA testing for Noonan syndrome is offered to the family. The child's parents are considering having their child tested for mutations in Noonan syndrome–associated genes. Which of the following considerations is the most appropriate to be used by the genetic counselor to help the family decide to proceed with the genetic testing in this patient?
    A. Confirming the diagnosis will not make the clinical situation worse.
    B. Confirming the suspected diagnosis may lead to appropriate changes in medical management.
    C. Testing will confirm paternity.
    D. Testing will improve the patient's insurability.
    E. The genetic test is cheap.

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