Genetic disorders impact not only the physical condition, but also the psychological and social health of patients and their families. Understanding the unique aspects of genetic information and anticipating reactions to genetic tests and diagnoses can help guide a course of action to minimize distress and maximize benefit for both the patient and family. Referrals to specialists or support services can also help address the psychological health of the patient and family.

The personal and permanent nature of genetic disease can raise a range of emotions including guilt, fear, and helplessness. Specialists such as genetic counselors, social workers, and psychologists, as well as members of support groups, can be extremely helpful to patients and families as they deal with these difficult issues.

Chapter 7 : Psychological and Social Implications
7.1 Genetic Information and Other Medical Information

Genetic information is often perceived as different from other medical information. Some people believe that genetic information is uniquely sensitive because of its predictive nature and potential implications for other family members; therefore, it raises unique social issues. This belief has translated to specific policy positions to protect genetic information and prevent it from becoming part of a patient’s medical record. Others believe that genetic information is like other medical information, and the same protections and high standards of privacy and confidentiality should apply to all personal medical information. In fact, they believe that treating genetic information differently from other medical information may result in unintended disparities.

Regardless, both genetic and non-genetic medical information:

- Have the potential to help or harm patients and must be considered in making patient care decisions
- Are complex and demands thoughtful, critical communication of risks and uncertainties
- Will arise in your practice; it is helpful to think through how you will respond in the face of inevitable questions, some of them involving difficult decisions

Concerns that may be specific to genetic information:

- Provides information about family members and relatives; disclosure of genetic information often directly impacts other family members
- Can lead to breaches of confidentiality that must be considered and addressed proactively

7.2 A Lifetime of Affected Relationships

Genetic conditions have powerful effects on families. Like many chronic conditions, they may require continuous attention and lack cures. They have implications for the health of relatives. So a genetic diagnosis for one family member may mean other biological relatives are at risk, even if they currently show no symptoms. In addition to the medical implications, genetic disorders present emotional challenges and special reproductive implications. Families may be concerned about difficult treatment options, the chance that additional offspring will inherit the condition, and prenatal and newborn testing decisions.
7.3 Impact of A Genetic Diagnosis

The psychosocial impact of a genetic disorder varies by the nature of the condition, the relationship of a person to the affected individual, and individual personalities. Every family is different, and it is difficult to predict how people will react to a genetic diagnosis. It is helpful to think in advance about some of the possible reactions so you can be prepared and minimize distress.

7.3.1 Patients. A genetic diagnosis can provide a great benefit to patients. When the condition is rare and patients and families spend years without knowing its name or cause, a diagnosis can help make sense of the situation. Diagnoses can lead to improved treatment options and access to support services. They can also help other family members make decisions about their own lives.

A genetic diagnosis may lead to negative reactions, too. A common response is that the science of genetics is confusing and frustrating. Patients identified with a genetic diagnosis may consider themselves at fault or “broken” or interpret their diagnosis as leading to something they cannot handle. A genetic diagnosis can lead to concerns about stigmatization.

The reaction to a diagnosis varies from individual to individual and is affected by many factors including age, gender, education, religion, and culture. Providers should be aware of these differences and understand the patient’s background in order to communicate effectively.

7.3.2 Parents. Understandably, diagnosis of a genetic condition may put stress on a relationship. Couples with an affected child often face difficult family-planning decisions because future children may have a chance of inheriting the condition. Depending on the condition, parents may also be faced with hard choices regarding prenatal testing and termination of a pregnancy. The magnitude of these decisions and their outcomes impacts both individuals and relationships. Parents may experience guilt due to the hereditary nature of genetic conditions.

7.3.3 Family. Given the shared nature of genetic information, it is important to consider the family unit. Unaffected family members should not be forgotten in the case of a genetic disorder. When one family member is diagnosed with a disease, family members who do not have the disease often feel guilt that loved ones are affected when they are not. For adult-onset diseases, unaffected spouses may view their partners differently. The diagnosis can lead to a breakdown in communication. Siblings of children with special needs sometimes feel neglected because parents may focus more time and effort on the siblings affected by a genetic condition. Including unaffected family members in the care of individuals with special needs can help them examine their own emotional issues. Adults who are diagnosed with a genetic condition and are considering having a child may need to consider the chance of having an affected child, as well as their ability to care for the child.

In cases where a genetic test is predictive, other family members may misinterpret the results as a diagnosis rather than an indicator of risk for a condition. It is important to keep in mind that genetic test results are often complex and may be difficult for patients and their families to understand. In some cases, a genetic test may reveal the risk status of other family members who may not wish to know this information, potentially encroaching upon their autonomy or privacy.
The financial burden of a chronic genetic condition can also lead to stress among family members. A family already struggling financially may be intimidated by the costs associated with caring for a child with special needs. Referrals to appropriate support services are crucial to help ease the stress caused by a genetic diagnosis. Advocacy groups, state health departments, and The Patient Advocate Foundation are all organizations that may provide a starting point for support services.

In general, support or advocacy groups and community resources can provide ongoing support to patients and their families with genetic conditions. Support groups provide a forum for sharing experiences about caring for a family member affected with a genetic condition, coping with a new diagnosis, obtaining healthcare or other services, and healing. Members of support groups know firsthand what it means to be faced with a diagnosis and need accurate, up-to-date information. Staying connected with their community helps individuals fight the feelings of isolation that often surround families living with a genetic condition.

7.3.4 Communities. Genetic testing can also affect the community at large. Genetics has been used in the past to stigmatize and discriminate along ethnic or racial lines, and underserved or underrepresented communities often view genetic research and services with distrust. They may feel that the results of a genetic test, including newborn screening, will be used to segregate their communities. These fears often work in combination with other difficulties, including availability of services and health insurance, communication, and cultural barriers, when navigating the medical system.

Some communities do not see their condition as a disability, but rather as one aspect of their lifestyle. For example, members of the deaf community may oppose hearing tests for this reason. In general, it is a good idea to understand the communities to which your patients belong so you can present information and options in ways that promote trust.

7.4 Coping Mechanisms

When a newborn is diagnosed with a genetic condition, parents may be overcome with concern for their child. Some common reactions include fear, confusion, and grief that their child is not “normal,” guilt that they did something to cause the condition, anger at the lack of a solution, or the belief that the other parent is to blame.

The fact that a medical cure or treatment may not exist often comes as a great surprise to parents. This further adds to the parents’ concerns about their ability to care for the child. The manner in which healthcare providers react has a large impact on how parents cope with negative feelings and can help them focus on the challenges and blessings of the newborn child.
The following suggestions can help healthcare providers help parents cope with the birth of a child with an inherited condition:

- Focus on the child’s overall well-being, not solely on the child’s genetic condition at routine visits. Talk about the newborn’s personality, feeding patterns, and other personal traits. Always remember that the newborn is an infant first and an infant with special needs second.

- Provide realistic expectations for the future and models for coping. The parents are likely to be asked many well-intentioned questions by relatives and friends, and parents will be better able to respond if they have thought about the questions themselves.

- Explain the genetics of the condition in an understandable manner. Consider referring the parents to a genetic specialist.

- Emphasize that you are aware of the difficulty of the situation and acknowledge that each parent has his/her own way of coping with the stress of caring for an infant with special needs. It may be helpful for families to share their feelings with others. Referrals to a social worker, psychologist, or support group may facilitate these discussions.

- Identify resources such as support groups that focus on the condition in question. Support groups can help families overcome feelings of isolation often associated with a rare genetic condition, provide firsthand experience about caring for an infant with the condition, provide information about expectations for the affected infant, and suggest coping mechanisms that will help both parents and siblings adjust to new challenges.

**Selected References**


Genetic Alliance
www.geneticalliance.org

National Organization for Rare Diseases
www.rarediseases.org

Organizations for Support Groups & Information (Genetic/Rare Conditions)
www.kumc.edu/geesupport/grouporg.html#specific

The Patient Advocate Foundation
www.patientadvocate.org