



CHAPTER 7 : PSYCHOLOGICAL & SOCIAL IMPLICATIONS

Genetic disorders impact not only the physical health, but also the psychological and social well-being 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 groups can also help address the psychological well-being of the patient and family.

An increased genetic risk or a genetic diagnosis can substantially impact medical management as well as the psychological and social well-being of the patient and family. The personal and permanent nature of genetic information raises a range of emotions including guilt, fear, and helplessness. Specialists such as genetic counselors, social workers, and psychologists, as well as support groups, can be extremely helpful to patients and families as they deal with these difficult issues.

7.1 GENETIC INFORMATION VS. OTHER MEDICAL INFORMATION

Genetic information, like other medical information:

- Has the potential to help or harm patients and must be considered in making patient care decisions.
- Is complex, demanding 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 judgments.

In addition, genetic information, unlike much of medicine:

- Provides information about family members and relatives. Disclosure of genetic information can often be helpful to family members.
- Can lead to breaches of confidentiality that must be considered and addressed proactively.

7.2 A LIFETIME OF AFFECTED RELATIONSHIPS

Genetic disorders have powerful effects on families. Like many chronic conditions, they may require continual attention and lack cures or treatments. They have implications for the health of relatives, so a genetic diagnosis for one family member may mean other blood relatives are also 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 the risk that additional offspring will inherit the condition, prenatal and newborn testing decisions, and difficult treatment options.



7.3 IMPACT OF A GENETIC DIAGNOSIS

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

7.3.1 Patients. A genetic diagnosis generally provides great benefit to patients. It helps patients understand their disorder, especially when the condition is rare and the patient has struggled to find a diagnosis. Oftentimes, patients spend years living with a condition without knowing its name or cause. Diagnoses usually 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. The science of genetics can be confusing, and patients are often frustrated until they understand the nature of their condition. Patients identified with a mutation may consider themselves at fault or “broken” or interpret their diagnoses as leading to something they cannot fight. A genetic diagnosis can lead to fears about insurance and employment discrimination.

The reaction to a diagnosis varies from individual to individual and is affected by many factors including gender, education, and religious and cultural beliefs. By being aware of these differences and understanding your patients' backgrounds, you will be able to communicate with your patients effectively.

7.3.2 Parents. Understandably, the diagnosis of a genetic condition may put stress on a relationship. For adult-onset diseases, unaffected spouses may view their partners differently, and the diagnosis can lead to a breakdown in communication. Couples with an affected child often face difficult family planning decisions because future children may be at higher risk. Depending on the condition, parents may also be faced with hard choices regarding prenatal testing and termination of pregnancy. The magnitude of these decisions and their outcomes has an impact on the individuals involved and on their relationship.

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 mutation, family members who do not have the mutation often feel guilt that loved ones are affected when they are not. Siblings of children with special needs sometimes feel neglected because parents need to focus more time and effort on their siblings. Including unaffected family members in the planning of care for individuals with special needs can help them come to grips with their own emotional issues. Adults who are diagnosed with a genetic condition and are considering having a child will need to consider the risk of having an affected child as well as their ability to care for the child.

In cases in which 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.

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 first-hand what it means to be faced with a diagnosis and to 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 or newborn screening will be used to segregate their communities. These fears often work in combination with other difficulties with the medical establishment, including communication and cultural barriers.

Members of the deaf community, for example, may oppose hearing tests for fear that deafness will be considered a disability rather than a lifestyle. In general, it is a good idea to understand the communities in 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 are 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, and 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. How care providers react makes a big 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 parents cope with the birth of a child with an inherited condition:

- For routine visits, focus on the child's well-being and not solely on the child's genetic condition. Talk about the newborn's personality, feeding patterns, and other personal traits and 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've asked the questions themselves already.
- Explain the genetics of the condition in an understandable manner and consider referring the parents to a genetics specialist, either a clinical geneticist, a genetic counselor, or genetics nurse.
- Emphasize that you are aware of the difficulty of the situation and acknowledge that each parent has his or her own way of coping with the stress of caring for an infant with medical needs. It may be helpful for families to share their feelings with others, and 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 first-hand experience about caring for an infant with the condition, provide information about expectations for the affected infant, and suggest coping mechanisms for both parents and siblings to adjust to new challenges.

SELECTED REFERENCES

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