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 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 issues and emotions including guilt, fear, and helplessness. Specialists such as genetic counselors, social workers, psychologists, or 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 family information can often be helpful to family members, but also 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 on the health of relatives, too—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 long-term medical implications of a disorder, genetic disorders present emotional challenges and special reproductive implications including the risk that additional offspring will inherit the condition, screening choices such as prenatal and newborn testing, and difficult treatment options.

7.3 Impact Levels

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 disorders, especially when the conditions are rare and the patients have struggled for a diagnosis. Oftentimes, patients spend years living with conditions without knowing their names or causes. 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.

How a patient reacts 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 better and be more effective.

7.3.2 Parents. The diagnosis of a genetic condition can have stressful effects 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 decisions related to family planning since future offspring may be at higher risk. Depending on the condition, parents may be faced with hard choices regarding prenatal testing and termination of pregnancy. The magnitude of these decisions and sense of loss has an impact on the individuals involved and on their interpersonal relationship. Also, a genetic disorder may create a situation where long-term caregiving is necessary. Those couples who are able to communicate their experience with others are most likely to move forward.

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 for a mutation, spouses, siblings, and parents 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 diagnosed with a genetic condition considering beginning or expanding their family will need to consider the risk of a having an affected child and/or their ability to care for the child.

In cases where a test is predictive, other family members may misinterpret the results of a genetic test as a diagnosis rather than an indicator of risk for a condition. It is important to keep in mind that results of a genetic test may be difficult to understand, not only for patients, but for their families too. In some cases, a genetic test may reveal the risk status of other family members who may not wish to know of this information, thereby encroaching upon others' autonomy or privacy.

The financial burden of a chronic genetic condition, too, can lead to stress among family members. If the family is already struggling financially, it may be intimidated by the costs associated with caring for a child with special needs. You can help your patients by referring them to such as the DC Health Families which ensures affordable health care for children, including doctor visits, prescription medicines, and transportation to appointments. Knowing that resources exist can 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 can help share experiences about caring for a family member affected with a genetic condition, how to cope with a new diagnoses, obtain healthcare or other services, and heal. Members of support groups know first-hand what it means to be faced with a diagnoses and to need accurate, up-to-date information, and helping individuals stay connected with their community helps fight the feelings of isolation that often surround families who may be living with a genetic condition.

7.3.4 Communities. Genetic testing can lead to issues for communities, not just individuals. 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/or services with distrust. They may feel that the results of a genetic test or newborn screening will be used segregate their communities. These fears are often in addition to more general issues with the medical establishment, including differences in communication styles and language and diverse cultural beliefs.

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 where your patients are from so you can present information and options in ways to promote trust and understanding.

7.4 Coping Mechanisms

When a newborn is first diagnosed with a congenital abnormality, parents are often 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 that there does not seem to be a solution, that God caused this, or that the other parent is to blame. The fact that a medical cure or treatment does not exist often comes as a great surprise to parents. This further adds to parents concern about their ability to care for the child in the future, too. 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.

Suggestions that can help parents cope with the birth of a child with an inherited condition include:

- For regular 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 a condition in an understandable manner and consider referring the parents to a genetics specialist—either a clinical geneticist, genetic counselor, or genetics nurse.
- Emphasize that you are aware of the difficulty of the situation and each parent has his or her own way of coping with the stress and caring for an infant with medical needs. It may be helpful for families to share their feelings with others and referrals to social workers, psychologists, or support groups may facilitate these discussions.
- Identify resources such as support groups which focus on the condition in question. Support groups can help families overcome feelings of isolation often associated with a rare congenital condition, provide first-hand experience about caring for an infant with that particular condition, provide information about expectations for the affected infant, and suggest coping mechanism for both parents and siblings to adjust to new challenges.

Selected References

Genetic Alliance *http://www.geneticalliance.org*

National Organization for Rare Diseases *http://www.nord.org*

Organizations for Support Groups & Information (Genetic/Rare Conditions) *http://www.kumc.edu/gec/support/grouporg.html#specific*

Burke W, Fryer-Edwards K, and Pinsky L E (eds). *Genetics in Primary Care (GPC): A Faculty Development Initiative.* 2001. *http://genes-r-us.uthscsa.edu/resources/genetics/pdfs/gpc-completedoc.pdf*