GENETIC COUNSELING: DOWN SYNDROME

Grand Canyon University

# Genetic Counseling: Down Syndrome

Modern genetics today establishes how diseases can be inherited based on DNA, genes and chromosomes. Down syndrome is a condition of genetic origin that is begun by the attendance of a complete chromosome - or part of a chromosome - in excess, specifically chromosome 21. Although Down syndrome is a genetic disease, it is not hereditary. As stated by the US National Down Syndrome Society (NDSS), Down syndrome affects one baby for 800 to 1,000 births (Natoli, et.al. 2012). It is important to know that the activities of the mother during pregnancy have nothing to do with Down syndrome. However, it has been established that the probability of having a baby with Down syndrome increases significantly when the mother is older or adolescent. Since some couples wait until they have children, the risk of having a baby by Down syndrome can increase. This is why parents with children in old age should get genetic counseling.

# Family History

The disease is responsive to genetic screening and prenatal diagnosis. The diversity of molecular analytical tests to identify a person or fetus congenital an illness that causes gene mutations. It can classify other family associates or families at danger of disorder or at the very danger of being a carrier. In antenatal, it leads to the difficult question of the medical termination of pregnancy, related to the history of each family, its convictions and beliefs. In neonatal, it is just as difficult (Skotko, et.al. 2009). Any announcement of a serious pathology must be done in a quiet place for listening and optimal availability. Adequate and fair information must be given to both parents about the disease, its natural evolution and its complications, its medical and social care.

It is in all cases a source of upheaval within families. It can lead to an emotional rejection of the child suffering from T21 (sometimes to an abandonment), to overprotection (to the detriment of the emotional support of the siblings), to parental tensions (depression, divorce), to a denial disability to come. A majority of children with T21 are also well inserted in their family, who has found the right balance and make happy this child "unlike any other”. The help of parents' associations can be helpful for these families (Bull, 2011).

# **Discuss the possible reactions to Counselling**

The possible reactions to counselling is that it is beneficial in helping patients recognize negative thought processes, begin to see themselves more realistically, and discover or unearth the reasons behind a negative self-image. While there is a debate about how to treat patients with poorer self-esteem, most professionals do agree that it is caused by internal thoughts and dialogues. Over time, if someone thinks or expresses negative things about himself, these thoughts will begin to reverse positive thoughts. This can lead to a bad, self-image, and often distorted (Assessment, 2000).

Those with very low self-esteem often do not consider themselves realistic. They may believe that they are physically taller than they are, or they may take small criticisms and setbacks to heart and internalize them. For example, someone cannot get a job they applied or they can be passed over for a promotion. Instead of thinking that maybe the position had already been filled or that it was just not a good fit, they start to believe that they were not chosen based on other factors. They may feel stupid and incompetent, or begin to believe that they can do nothing right.

These are distorted ideas based on the negative thoughts that manifest in the brain over time. Sometimes this pattern of self-harm is caused by an underlying emotional trauma. Most often, this trauma occurs in childhood, but it can also be related to a single traumatic event later in life. Counseling for down syndrome in these cases can be very beneficial because a counselor can help patients talk through and work past harmful issues in their lives.

Counseling is especially beneficial because trained counselors can help patients begin to re-evaluate how they perceive themselves. They can learn to recognize negative thoughts when they occur, and replace them with more positive thoughts. This can help brain recycle into possibly internalizing, and believe, positive thoughts. When using this tactic, it is important for patients to be honest and realistic. If a patient says he is the smartest person in the world, it could be harmful when he realizes that it is not true.

**Counseling the Patient**

The point of counseling for self-esteem is to allow patients to recognize and focus on their current strengths and stop weaknesses over-dramatize. A counselor not only knowing the proper techniques and mental exercises necessary to do this successfully, he or she will also provide a safe place for patients to talk and share their struggles and fears. Although self-esteem counseling cannot do all the work necessary to build a positive self-image, it can steer patients in the right direction.

**Conclusion**

With all this information, the geneticist arrives at a diagnosis and communicates to the family the characteristics of the disease, the role of genetic factors in its determination, the estimated prognosis for the consulting individuals and the possible treatments. The family is also informed about the possible risks of recurrence in future descendants or in other more distant members, such as cousins or uncles of the patient (Spahis, et.al. 1999).

Among the missions of the medical genetics team, a very important one is to offer people with a genetic disease and their families the opportunity to live and reproduce as best they can. The genetic diagnosis is fundamental to be able to approach the genetic counseling that is defined as a communication process about the human problems associated with the occurrence or risk of the occurrence of a genetic disorder in a family. It consists of explaining to the patient or his parents -according to the case-, the characteristics and causes of the disease that affects him, his different management alternatives, his prognosis and mode of inheritance, the risk of family recurrence and the possible reproductive options.

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