

GENETIC COUNSELLING

Evaluation of an individual with a genetic condition is not limited to the making of a diagnosis and the provision of medical treatment. Genetic counseling entails not only establishing the correct diagnosis and making recommendations for care, but also requires providing information to an affected individual or family regarding the risk of recurrence of a hereditary disorder. Genetic counseling is usually provided by a health care professional trained in Medical Genetics. This may be a doctor, a genetics associate, a Ph.D. medical geneticist, or other appropriate medical professional. Accurate counselling requires an accurate diagnosis and part of the medical genetics evaluation will include confirming the diagnosis and taking a family history.

The process of genetics counselling is, at least in part, one of education. Its purpose is to provide information to the individual and the family regarding diagnosis, use of molecular testing, management, future (prognosis), recurrence risk and reproductive options for the affected individual and family members. Genetic counselling also aims to help patients and families cope with their condition in the best way for them as individuals and to support families in doing this. This is an ongoing process and may require more than one visit.

Individuals with genetic conditions or parents of an affected individual are most likely to seek genetic counselling. However, this service may be requested by any relative or even by a prospective partner. Genetic disorders affect families and the need for counselling may extend to relatives outside of the nuclear family.

Genetic counselling may be appropriate for some families shortly after the birth of an affected child. However, detailed counselling may be overwhelming during this period, because the family may be more concerned about the health and survival of the newborn infant than about recurrence or reproductive risks. Counselling, in some instances, may not be appropriate until several months have passed after the birth of the affected child or may need to be repeated at different times as specific needs and questions develop over the lifetime of an individual.

Genetic counselling is an interactive process. Individuals and families will have questions that may not be anticipated by the Geneticist or doctor. Families may have questions which arise after genetic counselling has already taken place. Open lines of communication are vital and the individual seeking genetic counselling should not hesitate to return for follow-up appointments if questions or concerns remain.

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