

Genetic counseling

Genetic counselling is the process of advising individuals and families affected by or at risk of genetic disorders to help them understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. The process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- Education about inheritance, testing, management, prevention, resources
- Counseling to promote informed choices and adaptation to the risk or condition.

Professional roles of genetic counselors

Genetic counselors work in a wide variety of patient-facing and non patient-facing settings.

Clinical genetic counselors may provide general care, or specialize in one or more areas. For example: Prenatal and pre-conception, paediatrics, cancer, cardiovascular, neurology, assisted reproductive technology / infertility and psychiatry.

Outside the clinic, genetic counselors work in areas such as laboratories, research, education, public health settings, and corporate environments.

Counseling session structure

The goals of genetic counseling are to increase understanding of [genetic diseases](#), discuss disease management options, and explain the risks and benefits of testing. Counseling sessions focus on giving vital, unbiased information and non-directive assistance in the patient's decision-making process.

Attitudes toward genetic counseling and genetic testing

Many studies have examined the attitudes of the lay public toward genetic counseling and genetic testing. Barriers to obtaining genetic counseling include lack of understanding of genetics by both patients and healthcare providers, concerns about cost and insurance, and fears of stigma and/or discrimination.

Genetic Counselling in Pakistan

Monogenic hereditary diseases are diseases caused by defects in genes and currently it has been estimated that there are more than 10,000 monogenic diseases with a global prevalence of 10/1000 at birth.

According to estimation more than 350 million people are affected by monogenic genetic diseases globally. Thalassaemia, sickle cell anaemia, colour blindness, haemophilia, deafness, etc., are some examples of monogenic hereditary diseases. Pakistan is among the countries which

are on high alert for rapid increase or such genetic disorders. The reasons for high prevalence of hereditary disorder are many: one is consanguinity (blood relationship) due to cultural preference of cousin marriages and mate selection from near or distant blood relatives. Thus 80 percent consanguinity in Pakistan, is perhaps the highest in the world. The incidence of genetic diseases at birth can be reduced through introduction of premarital genetic counselling as a part of healthcare system. Genetic counselling and premarital genetic testing should be practiced in Pakistan to stop this rapidly increasing burden of hereditary diseases. In 2018, there are nearly 7000 genetic counselors practicing worldwide, across at least 28 countries.

Eugenics is a set of beliefs and practices that aim to improve the genetic quality of a human population by excluding certain genetic groups judged to be inferior, and promoting other genetic groups judged to be superior.