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# Medical Genetics: Clinical Practice and Treatment Options

Roberto Intilla*

Department of Medicine, Birla Institute of Technology and Science, Pilani, Rajasthan, India

Commentary

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***For Correspondence:** Roberto Intilla, Department of Medicine, Birla Institute of Technology and Science, Pilani, Rajasthan, India

**E-mail:** robertointilla@gmail.com

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## DESCRIPTION

Medical genetics is a branch of medicine that focuses on identifying and treating hereditary illnesses. Human genetics is a branch of science that may or may not have applications in medicine, whereas medical genetics refers to the use of genetics in the delivery of health care. Research into the causes and transmission of genetic illnesses, for instance, would fall under the authority of both human genetics and medical genetics, however the diagnosis, treatment, and counselling of those who have genetic disorders would fall under the scope of medical genetics. The study of normally non-medical phenotypes, such as the genetics of eye colour, on the other hand, would be seen as part of human genetics but not always pertinent to medical genetics (unless in cases like albinism). The phrase "genetic medicine," which is more recent, refers to medical genetics and encompasses topics like gene therapy, personalised medicine, and the quickly developing field of "predictive medicine."

Medical genetics covers a wide range of topics, such as the clinical work of doctors, genetic counsellors, and nutritionists, the use of clinical diagnostic laboratories, and studies into the aetiology and transmission of genetic illnesses. The application of medical genetics to numerous prevalent disorders is rising. As recent developments in genetics have revealed the aetiologies of morphologic, endocrine, cardiovascular, pulmonary, ophthalmology, renal, mental, and dermatologic disorders, overlaps with other medical specialities are starting to appear. Individuals who have undergone voluntary genetic and genomic testing are becoming more and more involved in the medical genetics field.

## Clinical practice

The range of practise, diagnostic procedures, and treatment strategies are all based on the clinical environment in which patients are examined. The following are examples of typical patient-genetic practitioner interactions for discussions:

- Referral to an in-hospital consultation or an outpatient genetics clinic, usually for a diagnostic assessment (pediatric, adult, or combination).
- Clinics specializing in genetics that treat lysosomal storage disorders, skeletal dysplasia, and inborn errors of metabolism.
- A suggestion for prenatal genetic counseling to go over pregnancy risks (advanced maternal age, teratogen exposure, family history of a genetic disease), test results (abnormal maternal serum screen, abnormal ultrasound), and/or prenatal diagnosis options (typically non-invasive prenatal screening, diagnostic amniocentesis, or chorionic villus sampling).
- Multidisciplinary specialty clinics (muscular dystrophy/neurodegenerative disorder clinics, cardiovascular genetics, craniofacial or cleft lip/palate, hearing loss clinics) that include a clinical geneticist or genetic counselor.

## Treatment

The DNA that makes up each cell in the body is contained in units called chromosomes. There is presently no medication that can reverse the genetic modifications in every cell of the body because genetic disorders are frequently brought on by changes to the chromosomes or genes. Therefore, there isn't a "cure" for hereditary illnesses at the moment. However, there are treatments available to control the symptoms of many hereditary disorders. When the disease's pathophysiology is known, as is the case, for example, with inborn metabolic abnormalities, nutritional and medicinal therapy may be used in order to prevent long-term effects. In other situations, the absence of the enzyme is replaced through infusion treatment. Recombinant enzyme infusions are used to treat some lysosomal storage illnesses because they can decrease the buildup of the chemicals in different tissues. Examples include Glycogen storage disease type II, Fabry disease, mucopolysaccharidoses, and Gaucher disease. Such treatments can occasionally cause allergic reactions and are constrained by the enzyme's capacity to reach the affected areas (the blood-brain barrier, for example, prevents the enzyme from reaching the brain). The therapeutic success of enzyme replacement therapy over the long term varies greatly between illnesses.