Genetic disorders delivering genetics service has certain challenges.

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Introduction

All diseases, to some extent, are influenced by genetics. Disease processes are influenced by variations in our DNA and variances in how that DNA operates (individually or in combination), as well as by the environment (which includes lifestyle). In this review, the genetic basis of human disease—including single gene disorders, chromosomal abnormalities, epigenetics, cancer, and complex disorders—is examined. It also considers how scientific knowledge and technological advancements can be used to provide patients with the best possible diagnosis, treatment, and care [1].

Finding DNA sequence variations that affect biological features, especially those involved in the beginning and course of human disease is one of the main objectives of human genetics. Technology advancements, basic genomic resources, analytical tools, and access to enormous volumes of genotype and phenotype data have all helped to accomplish this goal during the past 25 years. Genetic advancements have aided in the development of novel treatment and preventive approaches and greatly enhanced our understanding of the mechanisms underlying many common and unusual diseases. Delivering care that is customised to each patient's unique genetic predisposition patterns will be a growing focus of medical innovation [2].

Individual vulnerability is, to some extent, determined by genetic variation for practically all human diseases. As a result, evaluating the connection between sequence variation and illness propensity offers a potent tool for pinpointing key disease pathogenesis mechanisms and emphasising cutting-edge preventative and therapeutic approaches.

Many of the genes and variants that are responsible for rare diseases have been identified over the past 25 years thanks to advances in technology and analytical approaches, which frequently build on significant community projects, like those that produced the human genome sequence1 and elaborated on that reference to capture sites of genetic variation. These developments have also made it possible to systematically dissect the genetic basis of common multifactorial traits. The use of this knowledge to spur clinical care innovation is gaining momentum, most visibly through advancements in precision medicine. Previously limited to a few distinct clinical indications, genomic medicine is about to become widely used [3].

Since the discovery of DNA as the fundamental building block of heredity, the goal of medicine has been to be able to locally alter the human genome. The ability to improve genes through the correction of misplaced (mutated) genes or site-specific alterations with therapeutic treatment as the target is known as gene therapy. Following that, many tactics that are frequently employed for this objective are outlined. Gene therapy is now a field that primarily exists in research labs, and its application is still being tested. The majority of trials take place in the US, Europe, and Australia. It has the potential to treat acquired genetic diseases like cancer and some viral infections, including AIDS, as well as diseases brought on by recessive gene defects like Cystic Fibrosis, Haemophilia, Muscular Dystrophy, and Sickle Cell Anaemia [4].

Family history is frequently one of the strongest risk factors for prevalent disease complexes like cancer, Cardio Vascular Disease (CVD), diabetes, autoimmune disorders, and psychiatric illnesses, even though there are numerous potential causes of human disease. In addition to receiving an entire gene pool from each parent, a person also receives a wide range of cultural and socioeconomic experiences from their family. The likelihood of developing a disease is regarded to be well predicted by a person's family history since they most precisely reflect the particular genomic and environmental interactions that an individual encounters. There is no doubt that inherited genetic variation within families has a role in the pathophysiology of disease, both directly and indirectly [5].

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