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Recent Advances in Genetic Medicine

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Introduction

A diverse group of severe, early-onset conditions known as developmental and epileptic encephalopathies (DEEs) are characterized by developmental delay or regression, refractory seizures, and generally poor prognosis. Epilepsy affects nearly 70 out of every 100,000 children under the age of two, and genetic epilepsies account for 30% of all epilepsies and more than 0.4 percent of the general population. Epilepsy affects 5 8 million people a year in the United States, with an incidence of 35-71/100,000 per year. However, epidemiological data that are specific to DEEs are just beginning to emerge. One in 2,000 births were found to have severe epilepsy before 18 months, according to a larger study. Infantile spasms as well as the Dravet, Lennox–Gastaut, and West syndromes are among the DEEs that have received the most research.

Next-generation sequencing (NGS) has made significant progress in the study of human genetics and genomics over the past ten years, resulting in an explosion of gene discovery for a wide range of human diseases. Over 50 genes have been associated with epilepsy for the first time in the past three years alone, bringing the total number of disease-associated genes to 4,132. However, the brand-new technologies have also come with brand-new obstacles. "Phenotype expansions" associated with some disease genes are brought to light by the capacity to sequence a large number of affected individuals who share a variety of phenotypes but are nonetheless related to one another. Patients with epilepsy can present with static to degenerative clinical presentations, making it difficult to distinguish between isolated DEEs and secondary epilepsies associated with neurodevelopmental disorders (NDDs). The ability of NGS to provide a clinical diagnosis in a short amount of time is a significant advantage; however, the "cafeteria choice" of cutting-edge genetic tests that are readily available can confuse medical professionals and the families of patients [1].

Description

Many people's perceptions of the role and scope of genetic testing are changing as a result of advances in genetic technology's impact on the clinic. Genomic testing increases the likelihood of a diagnosis or prediction of a future diagnosis, but it also increases the likelihood of uncertain or unexpected findings, many of which may affect multiple family members. The increasing speed and availability of genomic testing is changing this, which means that genomic information is increasingly influencing decisions regarding patient care in the acute inpatient setting. In the past, genetic testing was rarely able to provide rapid results. The landscape of genetic conditions' treatment options is changing, which is changing clinical discussions about disorders that were once untreatable. In addition, the point of access to testing is changing as more services are offered directly to customers outside of the traditional healthcare setting. The ways in which genetic medicine is evolving in light of technological advancements are outlined in this review [2].

Case reports and genetic studies of the familial type are the most common, but there are few studies of the sporadic SHFM. A sporadic case of chromosomal duplication on 10q24.3 in a fetus is the focus of this case study. The study's objective is to discuss the underlying pathways and mechanisms that contribute to their development as well as provide clinical and molecular information about this abnormality. In addition, the study will suggest some prenatal diagnoses that can be used to plan molecular genetic tests to find mutations that cause diseases. The study will also emphasize the importance of genetic counseling, particularly in sporadic SHFM cases. This will be important to parents who decide whether or not to terminate a pregnancy because it will reduce the number of congenital and developmental abnormalities that occur at birth and ease the financial, psychological, and physical burden on the families affected [3-5].

Conclusion

The primary cause of genetic blindness is inherited retinal dystrophies (IRDs), which are characterized by the degeneration of photoreceptor and retinal pigment epithelial cells. The most prevalent IRD is retinitis pimentos, which has a worldwide prevalence of approximately 1/4000. Between 20% and 30% of patients with RP have an associated non-ocular condition, ranging from mild morphologic changes to life-threatening pathologies, in addition to the visual impairment effects. The genetic diversity of IRDs is staggering, with over 250 genes identified as causing syndromic and/or non-syndromic forms of the disease. Numerous IRD genes also exhibit phenotypic heterogeneity. Because of this, the clinical testing methods that are used to diagnose IRD electroretinography, optical coherence tomography, funduscopic examination, fundus auto fluorescence, dark adaptation, and visual field testing do not typically have the ability to predict the gene that is causing a patient's condition.

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Conflict of Interest

There are no conflicts of interest by author.

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