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Abstract Topic: - Clinical Genetics

Abstract Title: - Genetic Landscape of ALS: Significance of Advanced Couple Genetic testing

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Aims: - This abstract highlights Amyloid Lateral Sclerosis (ALS), a complex neurodegenerative disorder, which is inherited through an autosomal dominant pattern and also less frequently through an autosomal recessive pattern. This study also explores the significance of advanced couple genetic testing in individuals. This focuses on the impact of consanguineous marriages by identifying carrier status and assessing the risk of the disorder eventually promoting early detection and intervention.

Methods: - The genetic profile of an individual from a consanguineous marital background was analyzed by targeted carrier screening with whole exome sequencing (WES) on the Illumina sequencing platform, which is a cutting-edge genetic testing approach that provides a comprehensive analysis of the individual's risk status in connection to ALS.

Results: - The genetic screening of the individual revealed the occurrence of ALS during childhood. Early onset ALS, also known as juvenile ALS, is a less common but particularly concerning form of the disease. ALS typically occurs in individuals between 40 and 70, but in some cases, it can develop earlier. When it does occur in younger individuals, the progression and prognosis can vary widely, leading to severe disability. Life expectancy after diagnosis is between 2 and 5 years, but some people may survive for decades. The analysis also revealed a noteworthy association between consanguineous marriages and the incidence of amyotrophic lateral sclerosis.

Conclusions: - In conclusion, there is a heightened risk of ALS among individuals born to consanguineous unions, emphasizing the critical role of genetic factors in the pathogenesis of this disease. Given these findings, it is imperative to recognize the importance of genetic testing, particularly for couples in consanguineous relationships. Early genetic testing and counseling can help identify potential risks and guide individuals in making informed decisions about family planning. Such proactive measures can play a pivotal role in reducing the burden of ALS within consanguineous communities and, ultimately, contribute to the advancement of ALS prevention and treatment strategies.

Keywords: - The genetic screening of the individual revealed the occurrence of ALS during childhood. Early onset ALS, also known as juvenile ALS, is a less common but particularly concerning form of the disease. ALS typically occurs in individuals between 40 and 70, but in some cases, it can develop earlier. When it does occur in younger individuals, the progression and prognosis can vary widely, leading to severe disability. Life expectancy after diagnosis is between 2 and 5 years, but some people may survive for decades. The analysis also revealed a noteworthy association between consanguineous marriages and the incidence of amyotrophic lateral sclerosis.