Abstract ID: - 215

Abstract Topic: - Clinical Genetics

Abstract Title: - Copy number variation of SMN gene: Variability from Genotype to Phenotype

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Aims: - Spinal Muscular Atrophy (SMA) is a rare, autosomal recessive neuromuscular disorder characterized by the degeneration of motor neurons in the spinal cord, leading to muscle weakness and atrophy. This collection of case studies presents a comprehensive exploration of SMA, shedding light on the diversity of genetic mutations and clinical phenotypes observed in various patient populations with a particular focus on atypical and rare variants. We present few insights into the clinical spectrum of SMA by documenting a range of patient profiles, from neonates with severe Type 1 SMA to adults with milder forms of the disease; highlighting the crucial role of genotype-phenotype correlations in diagnosis and management.

Methods: - To investigate the spectrum of SMA types and SMN genotype distribution, we determined the copy numbers of SMN1 and SMN2 genes using the SALSA® MLPA® probemix P060-B2 SMA MLPA assay in Indian population.

Results: - We have determined total number of 51 cases, out of which SMA Type-1 with 43% (n=22), SMA Type-2 with 29% (n=15), SMA Type-3 with 6% (n=3) and other types with 10% (n=5); whereas, there are cases with 12% (n=6) having variable phenotype suggesting of spinal muscular atrophy but showed no copy number variation in SMN gene; suggesting the involvement of other point mutations or genes responsible for spinal muscular atrophy. Moreover, we explore the psychosocial and familial aspects of living with SMA, emphasizing the importance of multidisciplinary care and genetic counseling for affected individuals and their families.

Conclusions: - Collectively, by unraveling the genetic and clinical diversity of SMA to obtain the results which underscore the complexity of SMA, emphasizing the need for personalized approaches to diagnosis, treatment, and support. In sum, our study provides a holistic view of SMA, encompassing the diverse disease types and clinical presentations. Also, it enhances our understanding of different conditions and contributes to improved diagnostic and therapeutic strategies for individuals living with SMA across its various forms.

Keywords: - We have determined total number of 51 cases, out of which SMA Type-1 with 43% (n=22), SMA Type-2 with 29% (n=15), SMA Type-3 with 6% (n=3) and other types with 10% (n=5); whereas, there are cases with 12% (n=6) having variable phenotype suggesting of spinal muscular atrophy but showed no copy number variation in SMN gene; suggesting the involvement of other point mutations or genes responsible for spinal muscular atrophy. Moreover, we explore the psychosocial and familial aspects of living with SMA, emphasizing the importance of multidisciplinary care and genetic counseling for affected individuals and their families.