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Abstract Topic:- Complex traits and polygenic disorders

Abstract Title:- Classifying muscular dystrophy: Clinical presentations as harbinger of diagnosis and management

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Aims:-Since ancient times, clinical manifestations have been pivotal in diagnosing and managing anomalies. While recent advancements in genetic technologies have enhanced diagnostic capabilities, clinical observations in developing countries continue to be the benchmark of cost-effective diagnosis and treatment. Following this approach, our present study initially endeavors on classifying different forms of Muscular Dystrophy (MD).

Methods:- To do so, subjects were initially assessed based on various muscle specific symptoms like muscle weakness, enlarged calves, difficulties in walking and climbing. To delve further into their conditions, genetic testing was also employed to confirm the underlying mutations. Additionally, we integrated Ayurvedic principles to comprehend patient responses and provide personalized care, applying the ancient practice of "Prakriti" for present-time disease management.

Results:- Two individuals, a 10- and 14-years old male, exhibited mutations in the DMD gene, revealing Duchene MD, with deletions of exons 45 to 53. Intriguingly, despite sharing the same mutation, one maintained mobility while the other became non-ambulatory. In another case, a 20-year male displayed clinical symptoms with milder expression, ultimately leading to the diagnosis of Becker MD. A 30-year-old female presented with walking difficulties, muscle weakness, fatigue, and other symptoms, prompting an initial diagnosis of Limb-Girdle MD. Further, genetic analysis uncovered a homozygous variant in exon 2 of the POMGNT2 gene, indicating LGMD type 2 MD. Additionally, she was found to be a carrier of a heterozygous variant in the GNE gene, suggesting Nonaka myopathy. Two other subjects, aged 7 and 13, exhibited clinical symptoms such as frequent falls, calf hypertrophy, and joint contractures. Genetic testing revealed a point mutation in exon 8 of the COL12A1 gene, leading to a diagnosis of Bethlem Myopathy. Conversely, the second individual displayed a frameshift mutation in the COL6A3 gene, resulting in Ullrich Congenital MD type 2. Additionally, we found Kaphapradhaana-Pittaanubandhi to be the most prevalent constitution among MD patients, significantly shaping our treatment approach and emphasizing the holistic nature of healthcare.

Conclusions:- While molecular testing has offered in-depth insights into mutations, the complexities surrounding the reading frame, limited diagnostic capacities, financial constraints, and intricate genetic variations can make clinical observations a pragmatic and beneficial method. In conclusion, our study underscores the pivotal role of clinical observations in the initial diagnosis and phenotyping of MD conditions.

Keywords:- Muscular Dystrophy, Clinical Observations, Diagnosis, Prakriti, Bethlem Myopathy