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Abstract Title:- Prakriti and genetic profiling in Duchenne/Becker Muscular Dystrophy: a first ayurgenomic appraisal towards personalized care.

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Aims:-Nature and Nurture consonance is the only prospect to harvest eu-health. Duchenne Muscular Dystrophy (DMD; OMIM #310200) and Becker Muscular Dystrophy (BMD; OMIM #300376) are frequently found as severe aneu-health conditions. These X-linked recessive Muscular Dystrophies (MDs) stem from mutations in the dystrophin gene (DMD; OMIM #300377), encoding the dystrophin protein (Uniprot #P11532). The Prakriti concept, adjudged in Ayurveda, epitomizes an individual's nature-nurture and has gained significance in healthcare with the emerging field of Ayurgenomics. As current approaches offer only palliative option/s, the present research conducts a thorough analysis of demographic, clinical, genetic, and Prakriti profiles in the D/BMD Gujarati population by integrating ancient wisdom with modern paradigms.

Methods:- This study included 120 participants whose demographic and clinical data were assessed. The concentration of biomarker Creatine PhosphoKinase (CPK) was measured. Genetic analysis viz. MPCR, MLPA, and NGS were applied to identify aberration. Prakriti profiles of all participants were assessed using AyuSoft (C-DAC, Pune).

Results:- The demographic and clinical study highlights the heterogeneity in disease severity and progression with 15.83% of familial cases. The difference in CPK levels between DMD (12445.15±98.94 U/L) and BMD (8095±569.49 U/L) underscores the severity of these two types. Genetic analysis revealed 87.5% DMD and 12.5% BMD where 90% were identified as deletions, 4.17% duplications, and 5.83% point mutations. In the cohort, 84.96% of mutations were out-frame, and 15.04% in-frame. Deletion and duplication events were predominantly in distal regions (78.33%) and involved the central rod domain (65.83%). Among the detected deletions, 45-52 deletions exhibited dominantly in DMD subjects with heightened mutation frequency for exon 50. The study uniquely integrates Ayurvedic Prakriti profiles into consideration, contributing an additional layer of insight into the disease framework. Within our data, based on seven distinct Prakriti, three profiles emerged prominently. These are KaphpradhanaPitanubandhi (KP), KaphpradhanaVatanubandhi (KV), and VatpradhanaKaphanubandhi (VK). In both groups, 46.67% had KP Prakriti, whereas DMD had 40.95% KV and 12.38% VK, and 53.33% of BMD subjects had KV. The varying distribution of profiles between DMD and BMD introduces possibilities for Prakriti-based stratification of disease.

Conclusions:- Such integrative Ayurgenomics holds promise not only for a deeper comprehension of genetic conditions but also for paving the way for the development of innovative, real-time, and personalized management.

Keywords:- Duchenne Muscular Dystrophy, Prakriti, AyuSoft, Ayurgenomics, Management