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Abstract Topic: - Clinical Genetics

Abstract Title: - Evaluation and establishment of MMP9, TITIN and MYL3 as potential prognostic markers for Duchenne muscular dystrophy

Presenting author name: - Sheresha Khethavath

Presenting author institute: - Institute of genetics and hospital for genetic disease

Co-authors name: - Shilpa reddy.G, Sailaja.M, Manjusha.D, Pavani Ayyadevara, Vijaya L. Bodiga, , , ,

Co-authors institute: - Institute of genetics and hospital for genetic disease, Institute of genetics and hospital for genetic disease, Institute of genetics and hospital for genetic disease, Institute of genetics and hospital for genetic disease, Institute of genetics and hospital for genetic disease, , , ,

Aims: - Duchenne Muscular Dystrophy (DMD) is the most common neuromuscular X-linked disorders caused by mutations in the dystrophin gene consisting 79 exons, affecting 0.029 % live male births characterized by progressive muscle weakness and muscle degeneration. Reduced expression or absence of dystrophin protein leads to muscle cell necrosis; thus resulting in muscle weakness. Clinical Diagnosis of DMD is mainly based on the symptoms. Further Mutation spectrum is analyzed using MLPA technique. Creatine kinase (CK-M) is commonly used as a blood biomarker for DMD to evaluate the level of muscle damage and necrosis. However, it is not a reliable marker as it shows variation due to physical exercise, muscle injury, toxic agents etc. MMP9, TITIN, MYL3 are components of muscle membrane proteins that maintain the integrity of cell membrane. Abnormality in these proteins causes skeletal muscle damage and degeneration of muscle fibres. Recent studies suggest that loss of dystrophin or other components of DAPC results in leaking out of these proteins from muscle cell into circulating fluids. However very few studies are available in south Indian population. In this study, the qualitative analyses of these protein levels were done by immunoprecipitation / immunoblotting. Analysis was done in serum and urine samples collected from DMD patients with and without steroid treatment and were correlated with deletion and duplication status in the patients. Details of results will be presented. Variation in these protein levels in relation to the deletion and duplication status will help to understand the molecular Etiopathogenesis and to establish as prognostic markers for DMD.

Aim of the study is to evaluate and establish MMP9, TITIN and MYL3 as potential prognostic markers for Duchenne muscular dystrophy

Methods: - MLPA immunoprecipitation immunoblotting

Results: - Elevated levels of MMP9 ,TITIN and MYL3 are observed in DMD patients .

Conclusions: - MMP9, TITIN,MYL3 have great significance with disease progression. thus can be established as potential biomarker for DMD

Keywords: - Elevated levels of MMP9 ,TITIN and MYL3 are observed in DMD patients.