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Mutational Analysis in Dystrophin Gene with Dystrophinopathy: A Novel Familial Case Report in Tamil Nadu

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ABSTRACT Duchenne muscular dystrophy (DMD) is an X-linked neuromuscular degenerative disorder initiated by mutation in the dystrophin gene that is located on chromosome Xp21.The present case is a novel report of DMD with co-occurrence of Autism associated disorder, which has a similar genetic component. In this report, the researchers present a family based study of a 17 year old male who has been diagnosed with DMD. The objective of the present case report is to identify the genetic abnormalities of the DMD gene and associated neuro behavioral disabilities. The methodology of the study followed classical cytogenetic techniques in which genetic alterations showed deletion of exon 45 in chromosome Xp21.2. From this case study, the researchers report that, the mother is a carrier for transmitting DMD to her male offspring.