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

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Disorder

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.