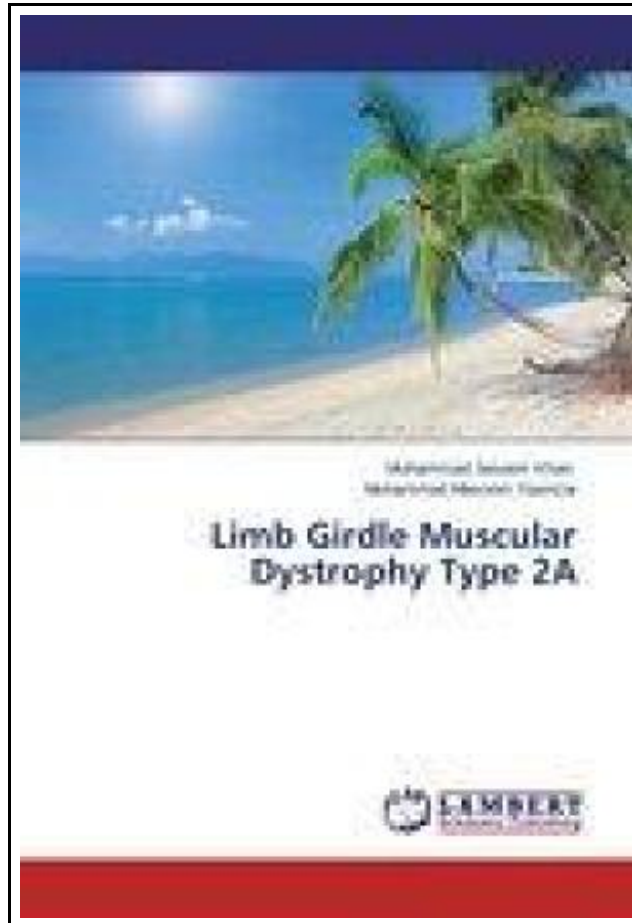


## Limb Girdle Muscular Dystrophy Type 2A



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LAP Lambert Academic Publishing Nov 2013, 2013. Taschenbuch. Book Condition: Neu. 220x150x4 mm. This item is printed on demand - Print on Demand Neuware - ABSTRACT Limb girdle muscular dystrophy (LGMD) is a heterogeneous genetically determined group of skeletal muscle disorders. Among the 24 reported subtypes of LGMD, limb- girdle muscular dystrophy type 2A (LGMD2A) is caused by mutations in the calpain 3 (CAPN3) gene. In Pakistan, consanguineous marriages are common due to social, ethnic and traditional customs which is a leading cause of calpainopathy. In the present study, two families (A, B) showing clinically distinct autosomal recessive limb girdle muscular dystrophy type 2A were evaluated genetically. Any other abnormality was not observed in both families. Technique of homozygosity mapping was used to track the gene responsible for autosomal recessive LGMD2A in both families. In homozygosity mapping with polymorphic microsatellite markers, linkage in family A and B with LGMD2A was established to CAPN3 gene on chromosome 15q15.1-q21.1. Subsequently, sequence analysis of all 24 exons of CAPN3 was performed but no disease causing DNA sequence variant was found in these exons suggesting the presence of mutation in the regulatory sequences or any other genes present in this region. 64 pp. Englisch.

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