



Approaches in Diagnostic & Care of Duchenne/Becker Muscular Dystrophy: Genotype-phenotype correlations in Duchenne/Becker muscular dystrophy patients

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LAP LAMBERT Academic Publishing, 2011. Taschenbuch. Book Condition: Neu. Neu Neuware; original eingeschweisst; Rechnung mit MwSt.; new item, still sealed; - This book presents a contemporary approach in the diagnosis, follow-up, treatment and care of Duchenne/Becker muscular dystrophy. Modern genetic tests are used to clarify molecular defects in our patients and to prepare them for future gene therapies. This is the first characterization of the cognitive impairment in Bulgarian patients with dystrophinopathy. The findings of the present study demonstrate that the cognitive profile of patients with dystrophinopathies is characterised by a broad range of intellectual abilities, as in addition to borderline general intellectual abilities, verbal short-term memory, is more severely affected than the long term one. A significant difference was found between general cognitive abilities in patients with an intact Dp140 isoform versus patients whose deletion was incompatible with formation of intact Dp140. 104 pp. Englisch.



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