GENOTYPE-PHENOTYPE CORRELATION OF COGNITIVE AND BEHAVIORAL CHANGES IN PATIENTS WITH DUCHENNE AND BECKER MUSCULAR DYSTROPHY

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Introduction: Duchenne and Becker muscular dystrophies (DMD/BMD) are caused by pathogenic variants in *DMD*, the dystrophin coding gene, and are characterized by progressive muscle weakness. However, intellectual disability and behavioral alterations have been reported since the first descriptions of these diseases. Recent studies associate the involvement of brain isoforms Dp140 and Dp71 with the development of intellectual disability. **Objectives:**To assess the cognitive and behavioral profile of patients with DMD/BMD and to associate these phenotypes with the location of the mutation in the *DMD* gene. **Methods:**Cross-sectional study with a convenience sample in which all patients followed up at the Medical Genetics Service of the HCPA with diagnosis of DMD/BMD aged ≥04 years were invited. Patients were evaluated for presence of Autism Spectrum Disorder (ASD), Attention Deficit Hyperactivity Disorder (ADHD) and Obsessive Compulsive Disorder (OCD) using specific scales applied by video calls. **Results:** Twenty- three patients and their guardians were interviewed. All patients were eligible for assessment using the CARS scale, of which 30.4% had a diagnosis of ASD. ADHD was present in 41.2%, and OCD in 20% of patients. CARS scores were higher among patients with variants that affect brain isoforms. There was no association between the location of the pathogenic variant with ADHD and OCD scores. **Conclusions:**Our study reinforces the association between the involvement of cerebral isoforms of dystrophin and the development of ASD in patients with DMD. Other factors must be sought to explain the presence of ADHD and OCD in the disease.

Key-worlds: Duchenne Muscular Dystrophy. Becker Muscular Dystrophy.Dystrophin Coding Gene.CARS scores.Intelectual disability.