P-043 - UNRAVELING CLINICAL PHENOTYPES IN SANFILIPPO SYNDROME TYPES A AND B: CLINICAL, BIOCHEMICAL AND NEUROIMAGING FINDINGS IN A COHORT OF BRAZILIAN PATIENTS

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INTRODUCTION: Mucopolysaccharidosis type III (MPS III; also known as Sanfilippo syndrome) belongs to a group of rare, genetic lysosomal storage disorders and is characterized by a deficiency in 1 of 4 enzymes involved in the degradation of heparan sulfate. Four subtypes of MPS III (A–D) have been identified based on the enzyme deficiency, along with their underlying genotypes and biochemical pathways. OBJECTIVES: To report 12 Brazilian MPS III patients (5 MPS IIIA and 7 MPS IIIB) and their “journey to diagnosis”. MATERIALS AND METHODS: Clinical, biochemical and radiological data retrospective analysis RESULTS: Symptoms generally began between the ages of 2 and 6 years and include developmental and language delays, hyperactivity unresponsive to medication, aggressive behavior and sleep disorders. All patients – but one - showed the typical progression of the disease, divided into 3 phases: phase I with developmental and language delay, frequent ear and respiratory infections, and diarrhea (not present in 4 patients who showed no apparent gastrointestinal features); phase II with progressive cognitive deterioration, behavioral difficulties, and sleep disturbances was present in all 12 patients; phase III with dementia, motor function decline, swallowing difficulties, and spasticity was seen in 10 patients (all of them older than 10 years of age) CONCLUSION: Patients with Sanfilippo can also present with symptoms that masquerade as a behavioral disorder. Several reports have shown patients to present with symptoms consistent with a variety of behavioral disorders, including autism, attention deficit disorder, and pervasive developmental disorder, which have resulted in misdiagnoses or delayed diagnosis of MPS III. All of our patients showed behavior problems in some point of the disease (attention deficit disorder and autism were the first diagnosis in 9 out of 12 patients). In our cohort, patients with MPS IIIA showed earlier onset of the disease, but no other clinical features that could differentiate it from MPS IIIB patients. Nevertheless, MPS IIIB patients showed wide range of clinical presentation with some early onset presentations (under 1 year of age) to the classical childhood presentation (2-4 years of age).