P-061 - PITFALLS IN THE RECOGNITION OF COGNITIVE PHENOTYPE IN PATIENTS WITH HUNTER SYNDROME (MPSII)

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Hunter syndrome (mucopolysaccharidosis II) is an X-linked disorder caused by the deficiency of iduronate-2-sulfatase. In this heterogeneous multisystemic chronic disorder, progressive and early cognitive decline occurs over time in 2/3 of the patients (severe or neuronopathic form of the disease) while cognition remains in the average range in the other third (attenuated or non neuronopathic form). The onset and the slowing of development is later and more variable than in MPSI and MPSIII starting at any age before 5 years old. **OBJECTIVE:** To create awareness about problems identifying progressive neuronopathic forms of MPSII even using adequate neurocognitive assessments. **METHODS:** As part of a prospective, longitudinal, observational study to evaluate neurodevelopmental status in pediatric patients with MPSII (HIT 090) we have screened 11 patients and recruited 6 male patients with formal cognitive assessments every 3 months in a median period of 2 years. The inclusion criteria were age range 2-13, sufficient auditory capacity and Developmental Quotient (DQ) >55 at baseline. The tools used were Differential Ability Scale (DAS II) for IQ and Vineland Adaptive Behaviour Scale (VABS II). **RESULTS:** In this small sample we have recognized 2 not related MPSII patients (ages 6 and 8 yo) previously categorized as having severe or neuronopathic form only by clinical impression but with borderline IQ and no cognitive decline during 2 years. Both had hyperactivity and language impairment probably secondary to early hypoacusia. One of them attended special school. By other hand, a 9 years old patient showed a stable low IQ (60-65) during the 18 months period of study but previous assessments from age 3 to 6 had shown a significant decline from DQ above 100. **CONCLUSION:** Identification of progressive neuronopathic phenotypes in MPSII is challenging. As new treatments are being developed for the neuronopathic form, to differentiate progressive from non progressive patients becomes crucial to evaluate efficacy of these treatments. A group of MPSII patients with mild behavioural-cognitive impairment but no declining over time (“pseudo-neuronopathic” form) should be identified as a minority phenotype.