P-047 - COEXISTENCE OF MUCOPOLYSACCHARIDOSIS TYPE IVA AND NEUROFIBROMATOSIS TYPE 1 IN A CHILD WITH A SEVERE PHENOTYPE

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INTRODUCTION: Mucopolysaccharidosis IVA (MPS IVA) is a genetic disease with an autosomal recessive inheritance. Prevalence ranged from 1 case per 71,000 to 1 per 500,000 live births. Neurofibromatosis type 1 (NF1) is a disease with an autosomal dominant inheritance that affects multiple organ systems and has a wide range of variable clinical manifestations. Average global prevalence is ~1 case per 3,000 individuals. OBJECTIVES: The aim of this report is to present the first case in the literature of the accidental coexistence of MPS IVA and NF1.

METHODOLOGY: Case report, it was approved by patient's family and the ethics committee.

RESULTS: The patient, now 4-year-old Colombian boy, is the third child of non-consanguineous healthy parents. He was vaginally delivered at 38 weeks of gestation. His birth weight was 3.4 kg (0.3 SD), length was 49 cm (-0.4 SD) and occipital frontal circumference was 42 cm (5.9 SD). He presented since his birth café-au-lait macules, skeletal bone dysplasia and swallowing disorder. He had a short stature (-3.6 SD) and low weight (-2.1 SD) at the age of 4 years. His medical history consisted of recurrent pneumonias (14 episodes), asthma and psychomotor retardation. Physical examination findings were macrocephaly, corneal clouding, tongue protrusion dystonia, dental fractures, pectus carinatum, generalized dysostosis, thoracic hyperkyphosis, giant melanocytic nevus and genu valgus. The patient have the pathogenic variant c.901G>T (GalNAc-6-sulfatase, GALNS, E.C.3.1.6.4) in homozygous state, which has a founding effect in our country, and the germline variant c.2326-1G>A (NF1 gene). A normal male karyotype (46, XY) was identified. Radiological findings showed bilateral hip dysplasia, platyspondyly and unfused posterior arch of C1. Enzyme replacement therapy was started at 2-years-old (elosulfase alfa, VIMIZIM®) with good clinical response and safety.

CONCLUSION: To our knowledge this is the first reported case of the coexistence of these two diseases, our hypothesis is that it was accidental, because they have different pathophysiologies. The patient has a severe phenotype that involves multiple organs and systems, taking into account that NF1 and MPS IV4 can affect the bone system. He had a good clinical response to enzymatic therapy however, his prognosis is poor.