P-089 - PERIPHERAL NERVOUS SYSTEM DISORDER IN NIEMANN PICK TYPE C DISEASE

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INTRODUCTION: Niemann Pick type C (NPC), a rare and progressive genetic disorder, is inherited in an autosomal recessive manner, with worldwide incidence estimated about 1/100,000 live births. In Brazil, until 2015, it were identified 75 new cases of NPC according to Rede NPC data. Pathogenic variants in the related genes must be found in individuals to confirm the diagnosis. The treatment is based on multiprofessional rehabilitation and the disease modifying therapy, Miglustat. OBJECTIVE: to describe six patients with NPC molecular diagnosis and clinical or electrophysiological peripheral neuropathy findings.

METHOD: A retrospective observational study of six individuals with infantile form of NPC based on biochemical criteria and/or molecular study, from 2002 until 2019, followed up in the Medical Genetics Service located in the Complexo Hospitalar Professor Edgard Santos – COM HUPES.

RESULTS: All the patients presented pathogenic variant at NPC1 gene, with diagnosis after at least 3 years of disease progression. Patient 1 presented juvenile form, and the others, early infantile form of NPC. All patients presented clinical signs of peripheral neuropathy, only 4 underwent electrophysiological study - ENMG. Two patients exhibited a demyelinating pattern and other two, axonal neuropathy.

DISCUSSION: Even-though peripheral nervous system commitment in NPC disease is still lacking of solid literature material, it has been published isolated case reports referring demyelinating peripheral neuropathy in infantile form patients. In this study, all patients presented evidence of peripheral neuropathy. The patient 1, juvenile form, exhibited axonal sensory-motor polyneuropathy, with polyphasic potential and reduced recruitment at ENMG. Patient 3, with five years of disease and two under specific treatment, presented axonal pattern, polyphasic potential and recruitment with severe rarefaction. The Patients 4 and 5, early infantile form, with 3 and 5 years of disease progression, respectively, prolonged latency and reduced velocity. They both showed a demyelinating pattern. Until 2003, 4 cases of NPC, infantile form, and peripheral neuropathy had been related in literature. In 2018, a patient with infantile form presenting pancytopenia and axonal peripheral neuropathy was published. Standard studies in individuals with NPC are needed in order to have adequate characterisation of peripheral nervous system commitment in the disease.