Acta Scientific Neurology (ISSN: 2582-1121)

Volume 5 Issue 4 April 2022

Editorial

Neuropsychology and Mucopolysaccharidosis

Nolly Nataly Castañeda*

Master's in clinical Neuropsychology (Cundinamarca), University of San Buenaventura: Bogota, Colombia

***Corresponding Author:** Nolly Nataly Castañeda, Master's in clinical Neuropsychology (Cundinamarca), University of San Buenaventura: Bogota, Colombia

Received: February 14, 2022

Published: March 01, 2022

Mucopolysaccharidosis is a rare or orphan disease of lysosomal storage, genetic, which is recognized by bone malformations, claw-shaped hands, macroglossia and enlargement of some organs such as the liver, spleen and heart. In South American countries this type of disease is unknown, however, the Colombian health system has recognized it within its legislation and has responded to the different demands for medical and psychosocial support for these patients and their families. But likewise, he faces neuropsychological needs from the family, social and school spheres, in which different studies are still being carried out.

In addition to the existence of different types of MPS (I, II, III, IV, VI, VII and IX) depending on the type of accumulated glycosaminoglycans, each with clinical subtypes determined by different enzymatic defects, show trigger serious and progressive clinical manifestations such as: exposed connective tissue, cartilages, bones and joints problems (MPS I, II, IV, VI and VII) and progressive and severe neurodegeneration for MPS types II, III and VII.

As a consequence of this enzyme deficiency, accumulation of glycosaminoglycans occurs in the cells of the body, causing multisystemic damage and revealing a characteristic phenotype for those who suffer from the disease; signs and symptoms that may range from the skeletal muscles, short stature, joint pain and stiffness, claw-like hands, prominent organs, affected systems including the respiratory tract, heart, viscera, corneal opacity, umbilical hernia and/or groin pain, carpal tunnel syndrome, hearing loss, thick lips and tongue, flat nose bridge and mouth, frequent throat infections, and "noisy" breathing (Maceira and Atienza, 2006). Manifestations in the central nervous system such as cognitive deficits, learning disabilities, spinal deformities and spinal cord compression (Cimaz., *et al*. 2009; Walker, 2013), even hydrocephalus, mental retardation and dementia (Pan., *et al*. 2009).

Studies related to brain damage in this type of disease are scarce, literature has reported behavioral and genetic studies in animals with MPS (Gagliardi and Bunnell, 2009), but in humans there is little scientific research reported, since, in general, the studies are limited to describing only the characteristics of the CNS found according to the type of MPS, as reported by Scarpa., *et al*. (2011).

Unfortunately for Colombia and other countries in South Asia, there are no epidemiological studies on the MPS, corroborated information or consolidated records on the population burden, as well as public health policies that respond to the needs of this population in a state of vulnerability in a comprehensive manner. There are no care and research networks that allow the monitoring and treatment of this type of pathology, as well as studies on the incidence, prevalence and natural course of MPS (Castañeda-Ibáñez and Sánchez-Campos, 2020), much less access to specialized care, advanced therapies, medications in general or neuropsychological profiles as such.

This situation is not alien to other scientific branches such as Psychology and Clinical Neuropsychology, but it is necessary to continue investigating and generating common knowledge in order to evaluate patients, create profiles that allow a better understanding of the disease and likewise proceed to other neurorehabilitation alternatives, which, as a purpose, would be one of the most significant contributions that could be made from neurosciences (Castañeda-Ibáñez and Acosta-Barreto, 2016).