LATM
LIFE SCIENCES CONSULTANTS

INsight

Mucopolysaccharidosis type II in Latin America

Mucopolysaccharidosis (MPS) is a group of genetic disorders caused by specific lysosomal enzyme deficiencies, leading to the accumulation of glycosaminoglycans (GAGs) – long chains of sugars (carbohydrates) – in cells and the extracellular matrix, and subsequently compromising tissues which leads to organ malfunction. Currently, MPS are categorized in seven clinical types (and numerous subtypes) in accordance with the specific lysosomal enzyme affected, leading to variable phenotypes, severity, and progression patterns among MPS types.

MPS type II (also called Hunter syndrome) is caused by a lack of the iduronate sulfatase enzyme. This clinical type is the only MPS disorder in which the mother alone can pass the defective gene to a son (called X-linked recessive). The disease is almost exclusively found in young males.

In Latin America, the MPS II landscape is different from the US and Europe. The incidence in Latin America oscillates between 0.69 and 1.19 cases in 100,000 live births. Furthermore, disease awareness in Latin America is low, and patient management as well as early diagnosis are still major challenges in the region. Although other countries in Latin America face large unmet needs and hurdles to address MPS II, Brazil is now a country of reference in the region due to their increased experience dealing with the disorder. A local KOL explained that, "*Casa Hunter and Genetics for All Institute are building a center that will enhance early diagnosis, development of genetic therapy and patient management by a multidisciplinary team*".

In Brazil, MPS II is the most common type, accounting for approximately 30-40% of the MPS disorders.

From 2000 to 2020, there have been 400 diagnosed patients in Brazil, with an incidence of 10-15 per year. In Colombia, MPS II is also the most frequent of the MPS disorders while in Argentina and Mexico that is not the case, where in Mexico the MPS type IV is the most common.

While the majority of MPS II cases are diagnosed in Brazil, in other Latin America countries this does not happen. An expert within the region mentioned that in Peru "*most of the cases are not diagnosed*", while in Chile "*there is a lack of specialized laboratories to perform the genetic test*", and in Colombia "*genetic test is very expensive*". He also mentioned that Brazil is an informal country of reference for diagnosing MPS II, stating that "*some Latin America countries send patient samples to Brazil in order to have the genetic testing performed*".

Although diagnosis of MPS II is a major problem in most Latin American countries, there are other hurdles to overcome. In some Latin America countries, there is no access to treatment. Another expert highlighted the lack of patient and family support, indicating that "*the family burden is very high. There is a need for weekly visits to the specialized center for the treatment. For those that reside far from specialized treatment centers the logistics and family financial impact is enormous*". In addition, treatment requires follow-up by a multidisciplinary, as this expert said that, "*treatment is usually coordinated by the geneticist, but patients suffer from multi-organ involvement and need a multidisciplinary team composed of neurologists, cardiologists, physiotherapists and other specialized healthcare providers*".

References

Giugliani, Roberto et al (2014), "Guidelines for diagnosis and treatment of Hunter Syndrome for clinicians in Latin America," Genetics and Molecular Biology, 37 (2), 315–29.

Khan, Shaukat A. et al (2017), "Epidemiology of mucopolysaccharidoses," Molecular Genetics and Metabolism, 121 (3), 227–40.

Office of Communications and Public Liaison (2019), "Mucopolysaccharidoses Fact Sheet," NIH Publication No. 19-NS-5115, (accessed March 10, 2021), [available at https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Mucopolysaccharidoses-Fact-Sheet].

LatM – Life Science Consultants has a key focus in Latin America
To know more about this topic please contact through:
https://latam.bio/

Miguel Ferreira
miguelferreira@latam.bio