Joual of Molecular Pathophysiology, 2022
VOL 11, NO. 7, PAGES 02

PERSPECTIVE

Open Access

Signs and Symptoms of Hunter Syndrome

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ARTICLE HISTORY
Received: 27-Jun-2022, Manuscript No. JMOLPAT-22-71024; Editor assigned: 30-Jun-2022, PreQC No: JMOLPAT-22-71024 (PQ); Reviewed: 15-Jul-2022, QC No: JMOLPAT-22-71024; Revised: 21-Jul-2022, Manuscript No: JMOLPAT-22-71024 (R); Published: 29-Jul-2022

Description

Large sugar molecules known as glycosaminoglycans accumulate in bodily tissues as a result of the rare genetic illness known as Hunter syndrome, also known as Mucopolysaccharidosis type II (MPS II). It is a lysosomal storage disease subtype. A lack of the lysosomal enzyme iduronate-2-sulfatase results in Hunter syndrome. Heparan sulphate and dermatan sulphate build up in all bodily tissues as a result of the lack of this enzyme. The only MPS condition with X-linked recessive inheritance is Hunter syndrome. The signs and symptoms of MPS I and Hunter syndrome are similar. It leads to problems in a variety of organs, such as the heart, bones, and respiratory system. In extreme situations, a teenager may pass away while still in their teens. Contrary to MPS I, this condition is not accompanied with corneal clouding.

Signs and symptoms

There are many different phenotypes that the Hunter syndrome can take. According to the presence of symptoms of the central nervous system, it has traditionally been classified as either "moderate" or "severe," however this is an oversimplification. Patients with "attenuated" or "mild" illness types may nonetheless experience serious health problems. The clinical course is often predictable for people who are seriously damaged; they typically pass away at a young age. A larger range of outcomes are available for persons with lesser types of the condition. Many people live into their 20s and 30s, but some people may enjoy nearly normal lives. Patients with milder types of the disease typically die from cardiac and respiratory problems.

In most cases, the signs of Hunter syndrome (MPS II) do not emerge at birth. Hernias in the stomach area, ear infections, runny noses, and colds are frequently among the initial signs. Signs of MPS II become more obvious as GAG accumulation in the body's cells continues. Many children with the condition look physically coarse, with high foreheads, flattened bridges on their noses, and swollen tongues among their facial traits. They could also have an enormous abdomen and a huge skull. Between the ages of 18 and 36 months, a diagnosis of severe MPS II is frequently made. A diagnosis is typically made between the ages of 4 and 8 years old in milder cases, where patients exhibit symptoms that are comparable to those of children with Hurler-Scheie syndrome.

Multiple organ systems become aberrant as a result of GAG accumulation. Children with severe MPS II may have skill loss and a reduction in development beyond 18 months. Heart function may gradually deteriorate as a result of thickening of the heart's walls and valves. Additionally, obstructive airway disease may develop if the walls of the airway thicken. Hernias may become more visible as the abdomen becomes bloated as the liver and spleen get bigger over time. By affecting all main joints, MPS II can cause stiffness and mobility restriction. A decline in the capacity to pick up little objects is caused by the finger and thumb joints becoming more and more involved. The impact on other joints, such the hips and knees, can make daily activities like walking more challenging. Hand function may further decline if carpal tunnel syndrome manifests. Short stature can occur as a result of bone problems. Some people with it may also have pebbly, ivory-colored skin lesions on their upper arms, legs, and upper back.

The condition is thought to be pathognomonic for certain skin lesions. Last but not least, the retention of GAGs in the brain might result in delayed development, intellectual impairment, and gradual function loss. In very young children, the ages at which symptoms first appear and the existence or absence of behavioural problems are indicators of the severity of the disease in the long run. Although the presence and severity of symptoms vary in each affected child, behavioural abnormalities frequently mimic combinations of signs of attention deficit hyperactivity disorder, autism, obsessive compulsive disorder, and/or sensory processing disorder. Aggression and a lack of a proper feeling of

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danger are frequently also present. The behavioural signs of MPS II typically appear before neurodegeneration and frequently get worse before the mental impairments get worse. Most children with severe MPS II have severe mental problems and are totally dependent on their caregivers by the time of death.

Pathophysiology

Critical bodily processes are supported by a wide variety of biochemical events in the body. The disintegration of big macromolecules is one of these processes. The fundamental issue with Hunter syndrome and associated storage problems is the failure of this mechanism.

The extracellular matrix, which is composed of various proteins and carbohydrates, is a component of the connective tissue and has a role in the biochemistry of Hunter syndrome. It assists in creating the body's structural skeleton. The matrix serves as the body's glue, encircling the cells in an ordered meshwork and holding them all together. A molecule by the name of proteoglycan makes up a portion of the extracellular matrix. Proteoglycans need to be broken down and replaced, just like many other parts of the body. Mucopolysaccharides are one byproducts of the body's breakdown of proteoglycans.