Hunter Syndrome

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Introduction

- Mucopolysaccharidosis II (MPS II) is an inherited disorder of carbohydrate metabolism that occurs almost exclusively in males, Characterized by distinctive facial features, a large head, hydrocephalus, enlargement of the liver and spleen (hepatosplenomegaly), umbilical or inguinal hernia, and hearing loss. Individuals with this condition may additionally have joint deformities and heart abnormalities involving the valves. (MPS II) is caused by mutations in the (IDS) gene and is inherited in an X-linked manner.[1]

Symptoms

- The signs and symptoms in Hunter syndrome is an enlarged head, large nose, large tongue, round and hard cheeks, thick lips, enlarging spleen and liver, rigid joints, recurrent respiratory tract infections, hydrocephalus, valve abnormalities and umbilical hernia.[1]

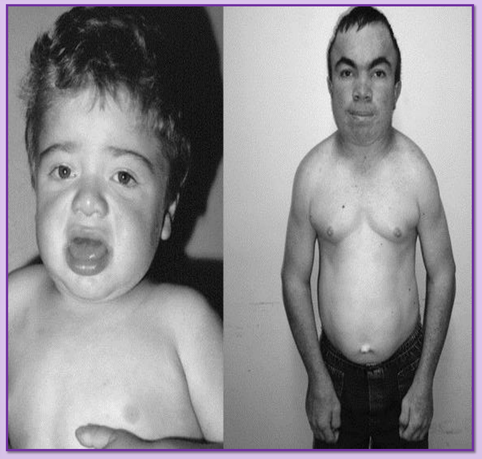

Figure 1 shows the facial changes in hunter syndrome.[1]

Conclusion

- Mucopolysaccharidosis type II (MPS II) is a rare X -linked recessive disorder in which the body is missing or does not have enough of an enzyme needed to break down long chains of sugar molecules, Without this enzyme, chains of sugar molecules build up in various body tissues and causing damage.

Causes

- Mucopolysaccharidosis II (MPS II) is caused by a deficiency of iduronate-2-sulfatase, which normally cleaves a sulfate group from the (GAGs), heparan and dermatan sulfate, Lack of (I2S) enzyme activity leads to an accumulation of (GAGs) within the lysosomes of various organs and tissues.
- The abnormal deposition of (GAGs) alters the architecture and function of cells and tissues, resulting in dysfunction of multiple organs and systems.[1]

Treatment

- Enzyme replacement therapy (ERT).
- Hematopoietic stem cell transplantation (HSCT).
- Substrate reduction therapy.
- Physical therapy can help with joint and movement issues. And occupational therapy can help you make changes at home and school to make it easier to get around and do things. [2]

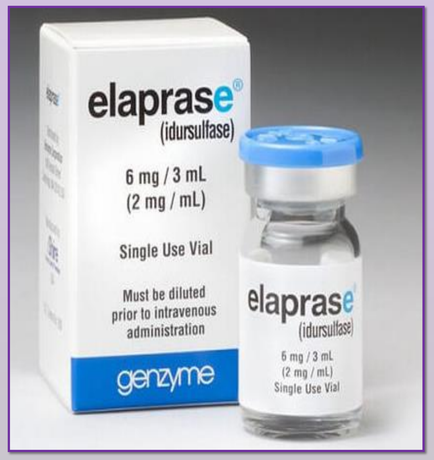

figure 2 shows the (ERT) drug. [2]

References

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2. Wraith JE. Mucopolysaccharidoses. In: Rimoin D, Korf B, eds. Emery and Rimoin's Principles and Practice of Medical Genetics. 6th ed. Philadelphia, PA: Elsevier; 2013:chap 102.