Mucopolysaccharidosis Type II

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Disease Overview

Summary

Mucopolysaccharidosis type II (MPS II) is a rare lysosomal inborn error of metabolism that affects every organ of the body. Although the age of onset, disease severity and the rate of progression of the disease vary significantly, initial symptoms and findings associated with MPS II usually become apparent in children from two to four years of age. Manifestations of MPS II may include not inflammatory joint stiffness, with associated restriction of movements; and coarsening of facial features, including thickening of the lips, tongue (macroglossia), and nostrils. Affected children may also have an abnormally large head (macrocephaly), a short neck and broad chest, delayed tooth eruption, progressive hearing loss, enlargement of the liver and spleen (hepatosplenomegaly), cardiac valve disease and progressive growth delays resulting in short stature. Two relatively distinct clinical forms of MPS II have been recognized. In the non-neuronopathic form (formerly defined as slowly progressive milder form) of the disease, intelligence

may be normal or only slightly impaired. In the neuronopathic form of the disease (formerly called early progressive more severe form), intellectual disabilities may be apparent in the early life of the patient.

MPS II is an X-linked genetic condition that mostly affects males; although a few females have been described as well, and is caused by changes (mutations) of the *IDS* gene that regulates the production of the iduronate 2-sulfatase enzyme. This enzyme is needed to break-down complex sugars, known as glycosaminoglycans, produced in the body.

Introduction

MPS II is one of a group of seven hereditary metabolic diseases known as the mucopolysaccharidoses, which in turn, are part of a group known as lysosomal storage disorders. Lysosomes function as the primary digestive units within cells. Enzymes within lysosomes break down or digest particular nutrients, such as certain carbohydrates and fats. In individuals with MPS disorders, including MPS II, deficiency or improper functioning of lysosomal enzymes leads to an abnormal accumulation of certain complex carbohydrates in cells (mucopolysaccharides, also known as glycosaminoglycans) within various tissues, such as the skeleton, joints, brain, spinal cord, heart, spleen, or liver.

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Programs & Resources

RareCare® Assistance Programs

Accepting Applications

Phone: 203-694-0412

Email: huntersyndrome@rarediseases.org Fax: 203-349-3190

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Additional Assistance Programs

Rare Disease Educational Support Program

Ensuring that patients and caregivers are armed with the tools they need to live their best lives while managing their rare condition is a vital part of NORD's mission.
https://rarediseases.org/patient-assistance-programs/rare-disease-educational-support/

Rare Caregiver Respite Program

This first-of-its-kind assistance program is designed for caregivers of a child or adult diagnosed with a rare disorder.
https://rarediseases.org/patient-assistance-programs/caregiver-respite/

Patient Organizations

Remember The Girls

NORD Member

Email: info@rememberthegirls.org

https://rarediseases.org/organizations/remember-the-girls/

National MPS Society

NORD Member

Email: info@mpssociety.org

https://rarediseases.org/organizations/national-mps-society/

Metabolic Support UK

Email: contact@metabolicsupportuk.org

https://rarediseases.org/organizations/metabolic-support-uk/

NIH/National Institute of Diabetes, Digestive & Kidney Diseases

Phone: 301-496-3583 Email: NDDIC@info.niddk.nih.gov

https://rarediseases.org/organizations/nih-national-institute-of-diabetes-digestive-kidney-diseases/

Society for Mucopolysaccharide Diseases

Email: mps@mpssociety.co.uk

https://rarediseases.org/organizations/society-for-mucopolysaccharide-diseases/

Canadian Society for Mucopolysaccharide and Related Diseases, Inc.

Email: info@mpssociety.ca

https://rarediseases.org/organizations/canadian-society-for-mucopolysaccharide-and-related-diseases-inc/

Let Them Hear Foundation

Phone: 650-462-3174 Email: info@letthemhear.org

Fax: 650-462-3144

https://rarediseases.org/organizations/let-them-hear-foundation/

Proyecto Pide un Deseo MÃ©xico, i.a.p.

Email: prayecto.pdeundese.mexico@gmail.com

https://rarediseases.org/organizations/proyecto-pide-un-deseo-maxico-i-a-p/

Xtraordinary Joy

https://rarediseases.org/organizations/xtraordinary-joy/

More Information

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