Mucopolysaccharidoses

Mucopolysaccharidoses consist of a group of inherited metabolic disorders. They are caused by a lack of the specific lysosomal enzymes. These are needed to break down mucopolysaccharides. Those are long chains of sugar molecules. These molecules are used to build connective tissues and organs in the body. When mutations occur in the genes for enzymes involved in the normal turnover of mucopolysaccharides, too many of them are stored in the body. This causes progressive damage. In most cases, it leads to eventual death. Eight clinical types and many subtypes of the disorders have been found. Each type differs clinically. But, in general, people with the disorder have a period of normal development. This is followed by a decline in physical and/or mental function. Some of the more common types include:

Mucopolysaccharidosis I(MPS I). This includes *Hurler, Scheie,* and *Hurler/Scheie syndromes.* Hurler syndrome is the most severe form of the disorder. It occurs in infancy. Symptoms include clouding of the cornea and progressive physical and mental disability. Death usually occurs before age 10. Patients with Scheie syndrome have milder symptoms and normal intelligence. Those with Hurler/Scheie syndrome suffer from intermediate symptoms.

MPS II is also called *Hunter syndrome.* It affects juveniles. It has a mild and a severe form. Signs of the severe form are:

- Joint stiffness.
- Mental deterioration.
- Dwarfing.
- Progressive deafness.

Death usually occurs by age 15.

Symptoms of the mild form include:

- Short stature.
- Limited motion.
- Features such as:
  - Enlarged forehead, lips, and tongue.
  - Misaligned teeth.

Life span may be normal.

Individuals with *MPS III,* or *Sanfilippo syndrome,* have progressive dementia and mental deterioration in childhood. Death usually occurs in the late teens.

MPS IV is also called *Morquio syndrome*. Symptoms usually appear in infancy. They may include severe dwarfing and corneal clouding. Intelligence is normal. Cardiac or respiratory disease may cause death in the third or fourth decade of life.

MPS VI is also called *Maroteauz-Lamy syndrome*. It resembles Hurler syndrome. Onset is in infancy. But intelligence is normal. Individuals may live into the second or third decade.

MPS VII is also called Sly disease. Symptoms include:

- Corneal clouding.
- Skeletal irregularities.
- Enlargement of the liver and spleen.

Intellectual impairments vary for this type of MPS.

DIAGNOSIS

Some of the excess mucopolysaccharides are excreted through the urine. So diagnosis often can be made through clinical examination and urine tests. Prenatal diagnosis is also available.

TREATMENT

There is no cure for these disorders. Treatment is symptomatic and supportive. In several individuals, bone marrow transplants have been attempted with varying degrees of success. Enzyme replacement trials are under investigation for MPS I and MPS II.

The prognosis varies depending on the type.

RESEARCH BEING DONE

The NINDS supports and conducts research aimed at finding the cause(s) of the various types of the mucopolysaccharidoses, and ways to treat, cure, and, ultimately, prevent the disorders. The genes for several forms of these disorders have been found. Scientists hope that understanding the genetics of the disorders may lead to breakthroughs in treatment.

Research is continually being done to learn more about this problem.

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