Hurler syndrome disease

Hurler's Syndrome

In a person's body every substance is important and should present the right amount of itself to function properly. In this case a very rare inherited disease of metabolism is when a person cannot break down long chains of sugar molecules called glycosaminoglycans. This disease is known as Hurler's Syndrome which is part of a larger group of diseases more commonly referred to as MPS. Other names for Hurler's Syndrome are alpha-L-iduronate, Mucopolysaccharidoss Type 1, and MPS1H. Other MPS diseases are Hurler Scheie Syndrome, which is a milder feature of Hurler Syndrome, Maroteaux-Lamy Syndrome, whose features are very similar to Hurler Syndrome and Sly Syndrome, whose phenotype is similar to that of Hurler Syndrome. Unfortunately, there are many more syndromes within the MPS disease category.

Hurler's Syndrome is named after Gertrud Hurler, who was the doctor that described both a boy and a girl with the condition in 1919. Dr. Scheie was a consultant ophthalmologist and in 1962 he wrote about some of his patients who were less severely affected than those previously diagnosed by Dr. Hurler. Those patients who could not be clearly diagnosed as either the severe or milder end of the disease were said to have Hurler Scheie Syndrome.

When you have Hurler's Syndrome, your body does not make a substance called lysosomal alpha-L-iduronidase. This means that a person without lysosomal alpha-L-iduronidase cannot break down long chains of sugar molecules which are used in the building of connective tissues in the body.

The sugar molecules are usually found in mucus and in fluid around a person's joints.

“ There is a continuous process in the body of replacing used materials and breaking them down for disposal.” What does break down long chains of sugar molecules is a substance called alpha-L-iduronidase enzyme. “ This is essential in cutting up the mucopolysaccharides called dermatan and heparin sulphate. The incompletely broken down mucopolysaccharides remain stored in cells in the body causing progressive damage.” A result of not having the alpha-L-iduronidase enzyme is that glycosaminoglycans will build up and damage a person's heart and organs. Symptoms of this disease can be mild or very severe.

Hurler's Syndrome is a disease inherited from both parents which means that both parents have to pass the gene to their child in order for the child to have the disease and any child of any race can have it. When a child is born you cannot really tell that they have the disease because they appear healthy at birth. “ Babies may show little sign of the disease, but as more and more cells become damaged, symptoms start to appear.” Most of the symptoms are thick coarse facial features with low nasal bridge, halted growth, progressive mental retardation, cloudy corneas, deafness, joint disease including stiffness, heart valve problems, abnormal bones in the spine, chronic runny nose, hernia, speech problems, hyper activity, depression, pain, and clawed hands. Most people do not notice the facial symptoms until the child is at the age of two. To tell if a person has Hurler's Syndrome, doctors usually do a simple urine test. The test would show extra mucopolysaccharides, but doctors would not be able to tell what form of MPS

that the person has. Thus, Hurler's Syndrome is not diagnosed until many other conditions have been looked into and after which more specific tests have been performed. This is usually done when the child is about six to twenty-four months of age.

To treat Hurler's Syndrome, a person would have to have enzyme replacement surgery which helps the body make alpha-L-iduronidase or have a bone marrow transplant to prevent retardation and that only improves some of the symptoms and should be done at a very young age. Scientists state that it is better for a child with the disease to have this surgery at a young age because they will have a better chance of living longer. A person can also have a stem cell transplant surgery and if this surgery is successful many symptoms of the disease are stopped. Seventy-five percent of those whose transplants are from siblings or other close family members have a higher rate of survival than those whose donors are not as closely matched. Many children with a heart disease caused by Hurler's Syndrome cannot have these surgeries due to the fact that their heart is not strong enough to help recover after any surgery. Any other treatments that can treat Hurler's Syndrome depend on the organs which are all infected. “ For those children whose diagnosis came too late in the progression of the disease to be eligible for transplant, there is still some hope in a newly approved enzyme replacement therapy.”

Young children with Hurler's Syndrome usually end up with nervous system problems and will have the chance of dying at a young age. Experts say that couples with the history of Hurler's Syndrome in their family or any other

disease should be tested before they think about having children of their own.

Today there is no actual cure for Hurler's Syndrome, but the MPS society is working very hard to find a cure for it. “ About 1 in 160 people are carriers, which result in about a 1 in 25, 000 chance of two carriers meeting. Since this disease is so rare the chance of having a partner who is another carrier is very slight provided their partner is not a cousin or other close family member.” Since Hurler's Syndrome is a recessive gene there are only 1 in 4 chances that a child will suffer this disease if their parents are carriers. In this case 1 out of 100, 000 children are affected with this rare disease. “ The unaffected children have a 2 in 3 chance of being carriers like their parents, and a 1 in 3 chance of being a normal non-carrier.”

The oldest survivor of Hurler's Syndrome is currently in his early twenties and is standing strong, however, most people affected with this disease do not live past the age of twelve.