Kentucky Newborn Screening Program, 275 East Main Street, Frankfort, KY 40621, Phone (502) 564-3756, Fax (502) 564-1510

Mucopolysaccharidosis Type I (MPS I) (Hurler, Hurler-Scheie and Scheie Syndromes)

CAUSE

Mucopolysaccharidosis type I (MPS I) is an inherited condition that affects many different parts of the body. It is a lysosomal storage disorder. When a lysosomal enzyme is missing or decreased significantly, the lysosome cannot break down certain substances. This causes a buildup of harmful compounds in cells throughout the body, resulting in a variety of symptoms.

SYMPTOMS

MPS I is a multisystem disorder and, depending on the amount of enzyme made by the body, can present at different times in life with a wide range of symptoms. The severe form, MPS I H, also known as Hurler syndrome, has more severe symptoms and usually starts within the first year of life. Symptoms of MPS I may include developmental delays and cognitive impairment or regression, short stature, stiff joints, speech and hearing impairment, heart and lung disease, enlarged liver and spleen, hernia, coarse facial features, hydrocephalus, spinal compression, pain and a shortened life span. The other subtypes of MPS I are MPS I H-S (Hurler-Scheie syndrome) and MPS I S (Scheie syndrome). Children with these forms of the disease may have normal intelligence with milder symptoms starting later in childhood but may still have many of the other symptoms that are seen in the severe type.

TREATMENT OPTIONS

Treatment of MPS I includes evaluation and management by a team of specialists who are knowledgeable about the disease. Many aspects of treatment are with symptomatic and supportive care based on the involved organs. Developmental, occupational, and physical therapy are often necessary. Treatment can include replacement of the missing enzyme through enzyme replacement therapy (ERT) to help relieve some of the symptoms but it is not a cure. Stem cell transplant is another available possible treatment for patients with severe MPS I.