KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

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

Pompe Disease

CAUSE

Pompe disease 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

The severity and age of onset of Pompe disease differ depending on the form.

- Classic infantile-onset form: Findings begin within a few months of birth, starting with heart problems, poor muscle tone (hypotonia), and muscle weakness (myopathy). Infants have failure to gain weight and grow at the expected rate (failure to thrive), feeding problems, and develop serious breathing problems.
- Non-classic infantile-onset form: Findings usually occur by 1 year of age and include delayed motor skills and progressive muscle weakness. The heart can be involved but not as severely as in the classic infantile form. Infants develop serious breathing problems.
- Late-onset form: Findings develop later in childhood, adolescence, or even adulthood and include progressive muscle weakness and breathing problems. It is less likely to involve the heart. Findings are usually milder and progress more slowly than in the other forms.

TREATMENT OPTIONS

Treatment includes replacement of the missing enzyme through enzyme replacement therapy (ERT). If a diagnosis of infantile Pompe disease is made, ERT should be started as soon as possible. ERT is recommended for the other forms as well but may be started later. Evaluation by genetics and additional medical specialists such as cardiologists, pulmonologists, and neurologists may be appropriate. Other supportive and symptomatic care is provided including therapies such as occupational and physical therapy.