INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Mucopolysaccharidosis Type 1H Newborn Screening Result

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from the heel of your baby and tested for rare, treatable diseases, including Mucopolysaccharidosis type 1H (MPS1H) or Hurler Disease.

What is Mucopolysaccharidosis type 1H (MPS1H)?
MPS1H is often called Hurler Disease and it is part of a group of a rare, inherited (genetic) diseases called lysosomal storage diseases. MPS1H happens when an enzyme called alpha-L-iduronidase (IDUA) is not working properly. IDUA helps to break down sugars in the body called glycosaminoglycans (GAGs) into smaller pieces. When the IDUA enzyme is not working, GAGs build up and cause problems in different parts of the body including the heart, brain, bones, eyes and ears.

MPS1H belongs to a group of diseases all caused by the deficiency of IDUA and together they are called MPS1. There are less severe (attenuated) forms of MPS1 called Hurler-Scheie disease (MPS1HS) and Scheie Disease (MPS1S). Babies with MPS1H usually have no (or very little) working IDUA enzyme, and those with MPS1HS or MPS1S may have some working IDUA enzyme.

What are the symptoms of MPS1H?
Babies with MPS1H often have no signs or symptoms at birth. Symptoms of MPS1H usually develop in the first year of life and tend to get worse quickly. Symptoms of MPS1HS and MPS1S usually start later in childhood or even adulthood, are milder than MPS1H, and generally do not involve the brain.

MPS1H symptoms can include:

- Developmental delay
- Thickening of the bones, and stiffness of the joints all over the body
- Large head, clouding of the eyes, and distinctive facial features
- Frequent ear infections and hearing loss
- Enlarged liver and spleen
- Hernia (soft out-pouching around the belly button or lower abdomen)
- Thickening of the muscles of the heart and heart valve problems

What does it mean if my baby has a positive MPS1H newborn screening result?
A positive MPS1H newborn screening result means that your baby may have a form of MPS1 and follow-up with a metabolic doctor is needed to know for sure.

What tests will help determine if my baby has MPS1H?
Blood and urine tests are usually done to find out if a baby has MPS1H, or one of the less severe forms of the disease. Sometimes, a small sample of skin may also be taken for testing. Your baby may also have a physical exam, x-rays, hearing tests, and other tests to look for signs of MPS1H.

When can my baby have these tests?
Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will contact you to talk about the results of your baby's positive newborn screen for MPS1H and arrange follow-up testing as soon as possible. If these tests show your baby may have MPS1H or another form of MPS1, quick follow up with a metabolic doctor will be arranged.

How is MPS1H treated?
Children with MPS1H are followed closely by a team of doctors and health care providers because many different parts of the body can be affected and symptoms can show up over time. There are two main ways to treat MPS1H in children. Both treatments involve getting IDUA enzyme into the body. One way to get working IDUA enzyme into the body is by bone marrow transplant (BMT, also called hematopoietic stem cell transplant). The other way is by enzyme replacement therapy (ERT).

Treatment for MPS1HS and MPS1S is usually not needed at an early age.

Why screen for MPS1H?
Screening helps find babies with MPS1H as early as possible so that treatment can be started sooner.

What happens when MPS1H is treated?
When treatment for MPS1H is started early, some symptoms can be prevented, and others can be improved or stabilize. Some symptoms of MPS1H can get worse over time and treatment can help to slow this down.

How does a baby get MPS1H?
MPS1H is an inherited (genetic) disease. Genes provide instructions for our bodies to make enzymes. The IDUA gene gives the body an instruction to make IDUA enzyme. A person with MPS1H (or one of the less severe forms of MPS1) has two copies of the IDUA gene that do not work. This means the IDUA enzyme that is made does not work properly. Each parent of a child with MPS1H usually has one non-working IDUA gene and is considered a "carrier". Carriers do not have MPS1H because they have a working copy of the gene that makes enough enzyme for them not to have the disease.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit our website at www.newbornscreening.on.ca

For more information about MPS1, please visit the Canadian MPS Society website at www.mpssociety.ca

NOTE TO PARENTS/GUARDIANS: This information is intended for parents whose baby has had a positive newborn screening result for Mucopolysaccharidosis type 1H (MPS1H). This fact sheet was written for information purposes only and should not replace professional medical advice, diagnosis, or treatment.