**Medical Provider Info for Mucopolysaccharidosis type 1 (MPS I)**

**(MPS I Carriers and Patients with Pseudo-deficiency alleles)**

**Introduction**: Mucopolysaccharidosis Type 1 (MPS I), is also historically and collectively known as Hurler syndrome, Hurler-Scheie syndrome, and Scheie syndrome. There is wide variability in severity and age of onset:

**2nd tier testing indicated your patient is a carrier, or has a pseudo-deficiency allele for MPS I**

## What is MPS I?

MPS I is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder (LSD). People with MPS I have lysosomes that cannot break down glycogen. This causes undigested sugar molecules and other harmful substances to build up in cells throughout the body, resulting in a variety of abnormal symptoms.

## MPS I Carriers

Carriers of MPS I are individuals who have a variation in one of their two IDUA (*alpha-L-iduronidase*) genes. These individuals still have one *IDUA* gene without a variation. **Carriers of MPS I do not have signs or symptoms of MPS I disease.**

However, there is a 1 in 4 (25%) chance a carrier may have a child with MPS I, if their partner is also a carrier of MPS I. Both parents of a child with MPS I are nearly always carriers of the condition.

## Pseudo-deficiency alleles

A pseudo-deficiency allele is a change in the body which results in lower IDUA enzyme activity, but not low enough to cause MPS I. **Patients with only a pseudo-deficiency allele do not have MPS I disease.**

## Pseudo-deficiency alleles in combination with an *IDUA* gene variant

Patients with a pseudo-deficiency allele **and** an *IDUA* gene variant most likely do not have MPS I disease either. But the patient may still need further evaluation with a metabolic geneticist to be certain.

## Where can I find more information?

Internet References: https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-i

http://www.babysfirsttest.org/