

Family action sheet for mucopolysaccharidosis type I (MPS I)

If a newborn blood screening result shows that your baby could have a serious condition called mucopolysaccharidosis type I (MPS I). This result requires further testing.

All babies born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is MPS I?

Mucopolysaccharidosis type I or MPS I can prevent the body from breaking down sugars properly.

This can affect many parts of the body, leading to bone, muscle, heart, and lung problems as well as developmental delays.

There are different forms of the disease, some more severe than others. The most severe form can appear in the first year of life. In a milder form, symptoms may not appear until later in childhood.

MPS I is an inherited condition. This means that it is passed from parents to children.

Is there treatment for MPS I?

Yes, there are treatments for MPS I. Early identification and treatment can help to delay the progression of the condition. Enzyme replacement therapy and sometimes stem cell transplant may be recommended as treatment. There are also new treatments being studied that may be available in the future.

Newborn screening takes blood from a newborn's heel to screen for medical conditions.



Next steps

1. Your baby's medical provider will refer you to specialists with MPS I experience.

The specialists will arrange for further evaluation to determine what treatment or monitoring may be needed for your baby.

2. Work closely with your baby's medical provider and specialists and follow their recommendations.

It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your infant's medical provider and specialists are the best people to teach you about MPS I. Find more information at these websites:

• [Baby's First Test](https://www.babysfirsttest.org)

(<https://www.babysfirsttest.org>). Type "MPS I" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español."

• [Health Resources & Services Administration \(HRSA\)](http://newbornscreening.hrsa.gov/conditions/mucopolysaccharidosis-type-I)

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