

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Mucopolysaccharidosis Type I (MPS I) to the Recommended Uniform Screening Panel (RUSP) in 2015.

About the condition

MPS I is a rare genetic disorder caused by a change in a single human gene. Studies of patients with symptoms suggest that about 1 out of every 100,000 people has MPS I. People with MPS I do not have enough of the IDUA enzyme that helps to break down certain waste products in cells. Babies with MPS I appear normal. There are 2 types of MPS I: the severe type and the attenuated type. Most children with MPS I have the severe type. In this type, MPS I can cause problems with the heart, airways, eyes and ears, muscles, bones, joints, and brain. These problems can worsen quickly and cause early death.

Treatment for MPS I

There is no cure for MPS I. Early diagnosis allows early monitoring and treatment for babies with MPS I. Treatments that can stop MPS I problems from getting worse include enzyme replacement therapy and human stem cell transplantation, also called a “bone marrow transplant.” The treatment a patient receives depends on many factors, including the type of MPS I.

Detecting MPS I in newborns

Newborn screening for MPS I can happen along with routine newborn screening for other conditions during the first few days of life. Newborn screening for MPS I measures IDUA enzyme activity. This process uses the same dried blood spots already collected for screening of other disorders. Newborns with low IDUA activity are at higher risk for MPS I. They need more testing and evaluation to diagnose the condition.

Public health impact

Based on what is known about screening and the risk of being born with MPS I, experts think that screening all newborns in the United States for MPS I would find about 44 babies with the condition each year (about 1.1 out of every 100,000 children born). It would prevent up to 2 deaths before age 5 years due to the disease each year.

Committee decision

The Committee voted in 2015 to recommend adding MPS I to the RUSP. As of 2016, the RUSP recommends that state newborn screening programs include MPS I.