

## EXECUTIVE SUMMARY

**This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Mucopolysaccharidosis Type II (MPS II) to the Recommended Uniform Screening Panel (RUSP) in 2022.**

### **About the condition**

MPS II is a rare genetic condition caused by changes in a single human gene. Studies of patients with symptoms suggest that fewer than 1 out of every 100,000 people in the United States (US) has MPS II. People with MPS II have low activity of the I2S enzyme that helps break down certain waste products in cells. Babies with MPS II look normal. There are 2 main types of MPS II: the severe type and the attenuated type. Both types of MPS II can cause problems with the liver, spleen, heart, airways, lungs, bones, joints, abdomen, head and neck, ears, mouth, nose, skin, throat, brain, movement, and behavior. People with the severe type of MPS II have more brain and behavior problems. Problems from MPS II can cause early death.

### **Treatment for MPS II**

There is no cure for MPS II. Early diagnosis allows early monitoring and treatment. Enzyme replacement therapy (ERT) is the most common treatment for MPS II. People with MPS II who get ERT have treatment for a few hours once a week at a hospital. ERT can slow down the disease process. It may also help people with both types of MPS II live longer.

### **Detecting MPS II in newborns**

Newborn screening for MPS II can be included with routine newborn screening for other conditions in the first few days of life. Newborn screening for MPS II measures I2S enzyme activity. This process uses the same dried blood spots already collected for screening of other conditions. Newborns with low I2S activity are at a higher risk for MPS II. They need more testing to know if they have MPS II and to find the right treatment.

### **Public health impact**

Experts used what is known about screening and the risk of being born with MPS II to assess the public health impact of screening. They think that screening all newborns in the US for MPS II would find about 59 babies with MPS II each year. This is the same as about 1.6 out of every 100,000 children born.

### **Committee decision**

The Committee voted in 2022 to recommend adding MPS II to the RUSP. As of August 2022, the RUSP recommends that state newborn screening programs include MPS II.