

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Hemoglobin H (HbH) Disease to the Recommended Uniform Screening Panel (RUSP) in 2010.

About the condition

HbH Disease is a disease that affects a protein called hemoglobin. Between 1–21 out of every 100,000 children gets a diagnosis of HbH Disease. People with HbH Disease have problems with key genes for making hemoglobin. Babies with HbH Disease look normal. There are 2 types of HbH Disease: deletional and nondeletional. Most children have the first type. HbH Disease can cause problems with making blood. These problems can affect many parts of the body and lead to death during young adulthood.

Treatment for HbH Disease

Hematopoietic stem cell transplant may cure HbH Disease. Other treatments that may stop symptoms from getting worse include blood transfusions, iron-lowering therapy, and folic acid. Whether and when a child needs treatment depends on many things, like the disease type.

Detecting HbH Disease in newborns

Newborn screening for HbH Disease can happen along with routine newborn screening for other conditions during the first few days of life. Newborn HbH Disease screening measures levels of one type of abnormal hemoglobin in the blood. This process uses the same dried blood spots collected to screen for other disorders. Newborns with high levels of this hemoglobin are at higher risk for HbH Disease. They need more tests to diagnose the disease.

Public health impact

Detailed data on how newborn HbH Disease screening affects public health were not available at the time of the report. However, newborn screening allows diagnosis and monitoring early in life. Without screening, diagnosis can take time after symptoms begin; some babies may never receive a diagnosis. Screening newborns for HbH Disease may prevent problems or delays in diagnosis.

Committee decision

The Committee voted in 2010 to recommend not adding HbH Disease to the RUSP. To add HbH Disease to the RUSP, experts need to know more about how many people have the disease. They also need to know how well early diagnosis and treatment work and how screening affects public health.