EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Guanidinoacetate methyltransferase (GAMT) deficiency to the Recommended Uniform Screening Panel (RUSP) in 2022.

About the condition

GAMT deficiency is a rare condition. It is caused by changes in the *GAMT* gene. Fewer than 1 out of every 100,000 people in the United States (US) have this condition. People with GAMT deficiency have too little creatine and too much guanidoacetate (GUAC). They have serious brain and muscle problems that get worse over time without treatment.

Treatment for GAMT deficiency

There is no cure for this condition. Early diagnosis allows early monitoring and treatment. The main treatment is supplements that are taken by mouth for life. Treatment can also include diet changes. Early treatment may help with or even prevent symptoms.

Detecting GAMT deficiency in newborns

Newborn screening for GAMT deficiency can be included with routine newborn screening for other conditions in the first few days of life. It measures the levels of creatine and GUAC. This process uses the same dried blood spots already taken for screening of other conditions. Newborns with low creatine and high GUAC levels are at a higher risk for GAMT deficiency. They need more testing to know if they have this condition and to find the right treatment.

Public health impact

Experts think that screening all newborns in the US would find about 7 babies with GAMT deficiency each year. This is fewer than 1 out of every 100,000 children born.

Committee decision

The Committee voted in 2022 to recommend adding GAMT deficiency to the RUSP. As of January 2023, the RUSP recommends that state newborn screening programs include GAMT deficiency.