

Positive Result:

Blood Spot Screen Result Notification



Elevation of C4 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C4 acylcarnitine.

What does this mean?

High levels of C4 acylcarnitine can indicate that your child has a metabolic disorder, such as isobutyryl-CoA dehydrogenase (IBD) deficiency. A positive result does not mean your baby has IBD deficiency or a different metabolic disorder, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a metabolic specialist will help arrange for more testing for baby. Your baby will also be seen by a metabolic specialist.

What is IBD Deficiency?

IBD deficiency is part of a group of disorders called amino acid disorders. Children with IBD deficiency are unable to break down proteins from the food we eat. This may lead to less energy for the body and cause the features of IBD deficiency in some people.

What health problems can cause it?

Most children with IBD deficiency have no signs or symptoms. In very rare cases, a child may have symptoms such as muscle weakness (hypotonia), poor feeding and/or difficulties growing and gaining weight (failure to thrive), developmental delays, a low number of red blood cells (anemia), heart problems (cardiomyopathy), and seizures.

Because IBD deficiency is rare and usually does not cause symptoms, it is difficult to know if the health problems reported in children with IBD deficiency are caused by this condition or if those health problems are unrelated to the condition. The medical community is still learning about IBD deficiency and its significance.

What treatment options are available?

Children with IBD deficiency may not need any treatment, especially if they are asymptomatic. A metabolic specialist may recommend dietary changes, avoiding fasting, and/or supplements.

Children with IBD deficiency should see their regular doctor and a doctor who specializes in metabolic disorders.

Resources



Scan here for more information about newborn screening in Minnesota or visit our website:
www.health.state.mn.us/people/newbornscreening

MedlinePlus:

www.medlineplus.gov/genetics

Save Babies Through Screening Foundation:

www.savebabies.org

Newborn Screening Info Center:

www.newbornscreening.hrsa.gov