

2-Methylbutyryl-CoA Dehydrogenase Deficiency SBCAD (2-methel-bu-ter-il-co-A dee-hi-drodge-in-ace de-fish-in-sea)

FAMILY FACT SHEET POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called SBCAD (ess-bee-cad).

A positive newborn screen does not mean your baby has SBCAD, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like SBCAD.

What is SBCAD?

SBCAD affects an enzyme needed to break down proteins from the food we eat so they can be used for energy and growth. In SBCAD, an enzyme used to break down proteins is missing or not working properly.

A person who has SBCAD doesn't have enough enzyme to break down protein containing isoleucine, so harmful toxins can build up in the body.

SBCAD is a disorder that is passed on, or inherited, from a child's mother and father. Because SBCAD is a genetic disease, family members are at risk of having SBCAD too, even if no one in the family has had it before.

What problems can SBCAD cause?

SBCAD is different for each child.

Most children who have SBCAD are of Hmong descent and have no health problems.

Infants with SBCAD from ethnic groups other than Hmong may have health problems. If SBCAD is not treated in these children, they might develop:

- Sleepiness
- Low blood sugar
- Developmental delay

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for SBCAD?

There are treatments for children who have health problems from SBCAD. Treatments for children with SBCAD can include:

- Diet low in isoleucine - a dietician will help you set up the best diet for your child
- Medications to help the body get rid of harmful toxins

Children with SBCAD should see their regular doctor, a doctor who specializes in SBCAD, and a dietician.

Children with SBCAD can benefit from prompt and careful treatment.

Resources for parents

Genetics Home Reference
<http://ghr.nlm.nih.gov>

Organic Acidemia Foundation
www.oaanews.org

MN Children & Youth with Special Health Needs
(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening