UPMC/CHILDREN'S HOSPITAL OF PITTSBURGH
INFORM / DEPARTMENT OF GENETICS
4401 PENN AVE, PITTSBURGH PA 15224
International Network for Fatty Acid Oxidation Research and Management

SCAD

Short chain acyl-CoA dehydrogenase (SCAD) deficiency is one of a group of inherited protein alterations that affect the body's ability to make energy from fats during stress, illness and fasting. When the body has exhausted its stores of available sugars, it must turn to fats to make energy. In each cell in the body, this efficient breakdown of fats takes place in a special sac-like bodies called mitochondria. Generating energy from fats is a multi-round process. In SCAD defects, the first step in the final round of the breakdown of fats is missing or reduced. Fortunately, this loss of function at the very last round means this defect usually has less impact than those in earlier rounds. Today in the United States, the majority of SCAD patients are identified right after birth by the expanded newborn screening program. For most infants in the United States, a blood spot for this newborn screening is obtained from the infant's heel before they go home from the birthing facility.

Signs and symptoms

By following infants with defective SCAD proteins, as identified from newborn screening, it was shown that most people with SCAD mutations never will show any specific symptoms. Earlier reported clinical findings have included episodes of intermittent metabolic acidosis, coma from elevated blood ammonia (hyperammonemic coma), neonatal acidosis with elevated muscle tone (hyperreflexia), multicore muscle breakdown (myopathy), and muscle fat storage with failure to thrive, and poor muscle tone (hypotonia) (276, 277). In contrast to other fat oxidation defects, SCAD deficiency does not cause low blood sugar (hypoglycemia) or low ketones (hypoketosis).

Diagnosis

The majority of SCAD defects are identified initially through newborn screening of an infant's blood spot by tandem mass spectrometry. Because they cannot perform the last round of chain-shortening of fats, these infants will accumulate two specific four- carbon fat products known as butrylcarnitine and ethylmalonic acid in blood and urine. The majority of these infants are not ill at diagnosis. Because pure SCAD alterations are considered benign (274-277, 281) many newborn screening programs no longer report infants with butrylcarnitine elevations, especially those with lesser changes.

Both butrylcarnitine and ethylmalonic acid can accumulate in other metabolic defects besides SCAD. As a result, infants or children who have symptoms similar to those of fat oxidation disorders, along with increased levels of these two metabolites and other accumulating metabolites may be referred for further study to rule out other metabolic syndromes.

Genetics

SCAD deficiency occurs when an individual inherits one change (mutation) in the gene for SCAD (*ACADS*) from each parent (autosomal recessive). There are two common SCAD alterations (625 G>A and 511 C>T) that change the active protein but leave it with sufficient activity to function without causing illness (278-280). Other rarer mutations usually cause more loss of protein function and more accumulation of butrylcarnitine and ethylmalonic acid, but still rarely cause illness. One rare mutation inherited with one of the common alterations usually results in an intermediate loss of activity.

Treatment

At present, SCAD deficiency by itself is considered a benign condition with no characteristic symptoms that need to be treated.

References

Click here for a list of references in the scientific literature

Ask an FAOD expert a question about SCAD. Please note that specific questions about your individual child's medical problems cannot be answered.