SOUTHWESTERN NEWS

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UT SOUTHWESTERN RESEARCHERS IDENTIFY ENZYME DEFICIENCY RESPONSIBLE FOR SOME SUDDEN INFANT DEATH CASES

DALLAS – August 27, 199 – The discovery by UT Southwestern Medical Center at Dallas researchers that an enzyme deficiency is responsible for some infant deaths attributed to Sudden Infant Death Syndrome (SIDS) could lead to new postnatal testing to identify those at risk.

"Our observations suggest that this deficiency is a severe disorder contributing to early infant death," said Dr. Michael J. Bennett, a professor of pathology and pediatrics at UT Southwestern and director of clinical chemistry at Children's Medical Center of Dallas. "The defect is a deficiency in the SCHAD-short-chain L-3-hydroxyacyl coenzyme A dehydrogenase - enzyme."

The findings were published in the July/August issue of *Pediatric and Developmental Pathology.*

The deficiency occurs during the breakdown of fatty acids derived from fat stores in the body. The body uses these fats as an energy source when its normal energy supply of glucose, which is converted into glycogen, is used up. In the event of fasting, infants tend to exhaust their limited glycogen supply quickly and begin using the stored fat, Bennett said.

The clinical consequences of SCHAD deficiency -- an inborn error of metabolism -- occur when fatty acids from the stored fats enter the liver and fail to generate energy or to produce ketones. Ketones are a vital energy source for the brain because it cannot use fatty acids, unlike the heart and skeletal muscle, which use fat directly.

"In these infants, fat can get into the liver, but it can't be metabolized and released," Bennett said. "Hence infants' brains starve from the deficit of ketones, and they become comatose."

The researchers examined 150 cases for this study. While only 2 percent were due to SCHAD deficiency, the researchers hope their findings will prevent at least a small portion of the unexpected fatalities previously described as SIDS cases by developing immediate

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postnatal testing that would identify infants with the enzyme deficiency. According to the U.S. National Center for Infectious Diseases Centers for Disease Control, SIDS is the leading cause of death of children 1 month to 1 year of age. In the United States, 5,000 to 6,000 infant deaths are attributed to SIDS each year.

Based on their results so far, the researchers advocate avoiding any circumstances that would induce a fast in an infant. When an infant ingests fewer calories than normally consumed, the liver will not produce the needed ketones, and glucose will be used up too quickly, precipitating a potentially fatal coma.

"The knowledge of this diagnosis, along with other fatty-acid metabolic defects, may be very important to the families of affected infants because of the genetic nature of these abnormalities," Bennett said. "The clinical features of these disorders may be at least partially controlled if the diagnosis is made early in life, possibly preventing deaths due to these disorders."

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