SBS, SIDS or Metabolic Disorder?:
A screening test to differentiate them

by Alan R. Yurko

Unexpected or unexplained deaths of infants are tragic. There is a trend for law enforcement, child protection agencies and the justice system to rush to judgment in these cases where any amount of suspicion is raised, thus further destroying an already grieving family. I describe a testing procedure for all interested persons to rule out metabolic disorders.

Three to six percent of all sudden unexpected infant deaths are from inherited disorders of fatty acid oxidation (FAO.) There are numerous types of these disorders. The use of electrospray tandem mass spectrometry for analysis of these disorders is cost effective and an important step for families, doctors, lawyers and medico-legal experts to take in investigating such serious tragedies. The most frequently detected metabolic disorders are medium chain and very long chain acyl-CoA dehydrogenase deficiencies, as well as glutaric acidemia type I and II deficiencies, among others.

A method for detecting these disorders of fatty acid and organic acid metabolism has been developed. It uses small amounts of blood, which is routinely obtained at autopsy. It is then applied to filter paper. Other biological fluids can be used as well, such as bile, vitreous fluid and urine.

For more information on this, you can contact:

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Source: Chace DH, et al, Electrospray Tandem Mass Spectrometry for Analysis of Acylcarnitines in Dried Postmortem Blood Specimens Collected at Autopsy from Infants with Unexplained Cause of Death; CLINICAL CHEMSTRY 2001:47(7) 1166-1182

-transcribed by Susan Kreider, RN

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