

February 20, 2013

RE: Newborn Screening for Adrenoleukodystrophy

To Whom It May Concern:

I am writing to express my strongest support for the implementation of newborn screening for adrenoleukodystrophy (ALD) a genetic disorder of fatty acid metabolism, which causes adrenal insufficiency, neurological deterioration and death. The majority of pediatric patients who die from the disease have the cerebral form presenting in mid to late childhood.

Hematopoietic stem cell transplantation with related and unrelated donors can prevent progression of the cerebral form ALD ***IF performed before the patient has manifested clinical symptoms of the disease***. Conversely, if transplantation is performed in symptomatic children, the disease usually progresses and the child either dies or stabilizes in a severely debilitated state.

Most families are unaware of the fact that they are at risk for having a child with ALD until a child in the family becomes symptomatic. Usually it's too late to help that index case. While subsequent pregnancies or births can be screened, this approach fails to rescue children who are the first case in their family. Newborn screening for ALD would identify these children before they manifested symptoms enabling them to have access to curative therapy with hematopoietic stem cell transplantation.

Another important reason to identify these children with newborn screening testing is because some will present with life-threatening problems due to adrenal crises which are secondary to adrenal insufficiency which also develops in babies and children with ALD. In many children adrenal insufficiency occurs before the onset of neurological symptoms. We have seen a child present in adrenal crisis as early as 6 months of age due to ALD. As adrenal crisis is also life-threatening, and preventable, it is a second compelling reason to screen newborns for ALD.

Over the course of my 20+ year career, I have seen hundreds of children with ALD who were diagnosed too late for treatment and who subsequently died months to years after devastating neurological deterioration. Newborn screening is an intervention that could have saved these children's lives.



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Pilot studies using Mass Spectroscopy for newborn screening for ALD have been shown to be effective and ready for additional testing. The next step would be implementation in a state in the U.S. with a large number of births. The state of Connecticut meets these criteria and could move forward to implement newborn screening for ALD.

Babies who screen positive would have to undergo mutation testing. If mutation testing was also positive, twice yearly screening of adrenal function (through blood tests) from birth and neurologic function (with brain MRI and developmental assessments, +/- BAER) from 3 years of age would allow for boys with ALD to be identified in the pre-symptomatic state.

In summary, I endorse, with the highest level of enthusiasm, the idea that the state of Connecticut under take, the task of adding newborn screening for ALD to their current panel. Please do not hesitate to contact me if I can provide additional information in support of this request.

Sincerely,



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Professor of Pathology  
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