



Standards of Care for Newly Diagnosed Babies with ALD as laid out by the University of Minnesota Children's Hospital's ALD Specialists:

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Once a baby boy has been identified to have the ALD gene mutation, it is imperative that the Metabolic Center where he has been referred provide a compassionate, vigilant and cohesive Standard of Care by his caregivers. This should include specialists in Pediatric Endocrinology, Neurology, a Genetic Counselor, Metabolic Doctor and Primary Care Physician. All need to communicate openly with each other.

The biological mother of the diagnosed child needs to be screened to determine if she passed the affected X chromosome to her son. If she is positive then all of her children and siblings need to be tested. If it is a baby girl who has been identified and the mother tests negative, then the father will need to be screened as well. All "at risk" family members will need to be screened.

To insure the best possible outcome for boys, they need to begin ACTH tests at the age of six months and every six months thereafter. Although it has not been studied, it is thought that the lack of accurate diagnosis of Addison's Disease and delayed treatment may influence the onset of the childhood cerebral disease (ccALD) in some patients, especially if they experience an Addisonian crisis. Stress dosing of hydrocortisone needs to be in place for those who are identified to have adrenal insufficiency to prevent such an event.

At 18 months the boys will need to have an MRI of the brain to refer to for future comparison. The MRI should be repeated every six months along with the ACTH test. Because it is very unusual for boys to develop the cerebral form of the disease before the age of three and the fact that they are still building myelin, it is unnecessary to screen before then. Two boys identified in New York were unnecessarily put through Bone Marrow Transplant before their first birthdays. We do not want to see that mistake repeated.

It has been proven that the MRI will reveal changes before they are symptomatic which will enable them to undergo Bone Marrow Transplant giving them the best possible outcome afterward. Therefore, the MRI needs to be repeated every 6 months along with the ACTH tests through the age of 12. If they do not develop ccALD then the MRI's only need to be done annually from then on. It is highly recommended that all boys be referred for a basic work up at the University of Minnesota at age 6 months in order to keep a data base and track the outcome of ALD boys identified through newborn screening. They also offer their services to review the bi-annual MRI's as they have sometimes been able to identify changes not obvious to a radiologist who is not experienced with ALD.

Through the implementation of newborn screening and follow through with the Standards of Care, you will save many boys and prevent their family from having to experience the heartbreak and devastation that late diagnosis often brings. We need to empower the parents and make sure they realize that early diagnosis is a gift, not a death sentence. Despite the diagnosis of ALD, they should be able to enjoy their little bundle of joy, knowing that they are in the best hands and will receive the most professional care available.

Please pass on the brochure that was prepared for them by other families who have been through this journey, which also includes information about the many advocacy organizations that exist to provide support and love along the way.

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