

PRIMARY ADRENAL INSUFFICIENCY IN YOUNG BOYS IS A RED FLAG FOR ALD

- Adrenoleukodystrophy (ALD) is a frequent cause of primary adrenal insufficiency in young boys who present in the absence of adrenal antibodies¹
- ALD is a rare X-linked disorder resulting from mutations in the *ABCD1* gene.¹ The early symptoms of ALD are similar to other medical conditions, making diagnosis challenging¹
- ALD may develop into cerebral ALD, a severe form of the disease¹
- Cerebral ALD leads to severe neurodegeneration and often death within a decade of diagnosis²

SOME FACTS ABOUT ALD

- 85% of young boys diagnosed with ALD also had primary adrenal insufficiency³
- ALD occurs in about 1 in 21,000 males⁴
- Causes impaired peroxisomal β -oxidation of VLCFA, resulting in accumulation of very long chain fatty acids (VLCFA) in plasma and all tissues¹
- Clinical manifestation of ALD varies widely, even among members of the same family. Patients may exhibit any or all of a spectrum of phenotypes, including adrenal insufficiency, adrenomyeloneuropathy (AMN) affecting the spinal cord and peripheral nervous system in adults, and/or the rapidly progressive cerebral form (cerebral ALD)¹
- Early symptoms are variable and non-specific but may include behavioral or learning issues and audiovisual impairment¹
- About 35-40% of boys with ALD will develop cerebral ALD⁵ which leads to progressive behavioral, cognitive, and neurologic deficits and, in most patients, total disability and death within a decade of diagnosis²
- Symptoms generally present between ages 3 to 10⁵

MEASUREMENT OF VERY-LONG CHAIN FATTY ACID (VLCFA) LEVELS IN PLASMA MAY HELP DIAGNOSE ALD

- Increased plasma levels of VLCFA are suggestive of ALD¹
- Boys with primary adrenal insufficiency who are negative for 21-hydroxylase antibody should be assessed for ALD by measuring VLCFA levels in plasma⁶
- If you suspect ALD, refer the patient to a pediatric neurologist

EARLY DIAGNOSIS OF ALD COULD SAVE A LIFE

- Cerebral ALD can be successfully treated with hematopoietic stem cell transplantation, but only if the disease is diagnosed in its early stages⁷
- Early diagnosis gives boys with cerebral ALD the best chance at positive long-term outcomes



WHEN YOU SEE ANTIBODY-NEGATIVE PRIMARY ADRENAL INSUFFICIENCY—CHECK FOR ALD

References: 1. Engelen M, et al. *Orphanet J Rare Dis* 2012;7:51-64. 2. Mahmood A, et al. *Lancet Neurol* 2007;6:687-692. 3. Mahmood A, et al. *Pediatr Transplant* 2005;9 (Suppl 7):55-62. 4. Bezman L, et al. *Ann Neurol* 2001;49:512-517. 5. Moser HW, et al. *Nat Clin Pract Neurol* 2007;3:140-151. 6. Bornstein SR, et al. *J Clin Endocrinol Metab* 2016;101:364-389. 7. Miller WP, et al. *Blood* 2011;118:1171-1178.

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