



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

- The majority of cases of antibody-negative primary adrenal insufficiency in young boys are caused by adrenoleukodystrophy (ALD)<sup>1-3</sup>
- ALD may develop into cerebral ALD (CALD), a severe form of the disease<sup>4,5</sup>
- If untreated, CALD often leads to severe neurodegeneration and sometimes death within 2 to 3 years<sup>5</sup>

## EARLY DIAGNOSIS OF ALD COULD SAVE A LIFE

- If CALD is caught early, lifesaving treatment is possible<sup>6</sup>
- *Early detection is critical*

## CHECK VERY LONG CHAIN FATTY ACID LEVELS (VLCFA)

- A simple VLCFA blood test can diagnose ALD<sup>4</sup>
- *See the back of this card for more information*

### SOME FACTS ABOUT ALD

- 85% of young boys diagnosed with ALD also had primary adrenal insufficiency<sup>7</sup>
- Occurs in about 1 in 20,000 males<sup>3</sup>
- Causes impaired peroxisomal  $\beta$ -oxidation of VLCFA, resulting in accumulation of VLCFA in plasma and all tissues<sup>3</sup>
- Early symptoms are variable and non-specific but may include behavioral or learning issues and audiovisual impairment<sup>3,4</sup>

## ABOUT 35-40% OF ALD CASES WILL CONVERT TO CALD<sup>4</sup>

- CALD leads to progressive behavioral, cognitive, and neurologic deficits and, in most patients, total disability and death within 2 to 3 years<sup>4,5</sup>
  - Symptoms generally present between ages 4 and 8<sup>4</sup>
- CALD can be successfully treated with allogeneic haematopoietic cell transplantation, **but only if diagnosed in its earliest stages**<sup>6</sup>

## A SIMPLE BLOOD TEST CAN MAKE ALL THE DIFFERENCE

- Boys with antibody-negative primary adrenal insufficiency who test negative for 21-hydroxylase antibody should be assessed for ALD by a VLCFA assay<sup>8</sup>

VLCFA Assay	
<b>CPT code</b>	82726
<b>Preferred specimen</b>	1 mL plasma collected in an EDTA (lavender-top) tube
<b>Collection instructions</b>	Fasting sample required
<b>Transport temperature</b>	Frozen
<b>Specimen stability</b>	14 days (frozen)
<b>Methodology</b>	Gas chromatography–mass spectrometry (GC-MS)

Codes and information accurate for Quest Diagnostics screening laboratory.

## THE VLCFA ASSAY CAN DIAGNOSE ALD IN MOST AFFECTED BOYS

- If you suspect ALD, order a VLCFA test and refer the patient to a pediatric neurologist



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

Visit [[microsite URL](#)] for more information about primary adrenal insufficiency and its relationship to ALD, brought to you by bluebird bio.

Additional information on ALD and its symptoms and treatment is available at [ALDConnect.org](#).

**References:** 1. Lombard-Platet G, Savary S, Sarde CO, Mandel JL, Chimini G. A close relative of the adrenoleukodystrophy (ALD) gene codes for a peroxisomal protein with a specific expression pattern. *Proc Natl Acad Sci USA*. 1996;93(3):1265-1269. 2. Ronghe MD, Barton J, Jardine PE, et al. The importance of testing for adrenoleukodystrophy in males with idiopathic Addison's disease. *Arch Dis Child*. 2002;86(3):185-189. 3. Moser HW, Mahmood A, Raymond GV. X-linked adrenoleukodystrophy. *Nat Clin Pract Neurol*. 2007;3(3):140-151. 4. Steinberg SJ, Moser AB, Raymond GV. X-linked adrenoleukodystrophy. In: Pagon RA, Adam MP, Ardinger HH, et al, eds. *GeneReviews*<sup>®</sup> [Internet]. Seattle, WA: University of Washington; 2015. 5. de Beer M, Engelen M, van Geel BM. Frequent occurrence of cerebral demyelination in adrenomyeloneuropathy. *Neurology*. 2014;83(24):2227-2231. 6. Mahmood A, Raymond GV, Dubey P, Peters C, Moser HW. Survival analysis of haematopoietic cell transplantation for childhood cerebral X-linked adrenoleukodystrophy: a comparison study. *Lancet Neurol*. 2007;6(8):687-692. 7. Mahmood A, Dubey P, Moser HW, Moser A. X-linked adrenoleukodystrophy: therapeutic approaches to distinct phenotypes. *Pediatr Transplant*. 2005;9(Suppl 7):55-62. 8. Brett EM, Auchus RJ. Genetic forms of adrenal insufficiency. *Endocr Pract*. 2015;21(4):395-399.

