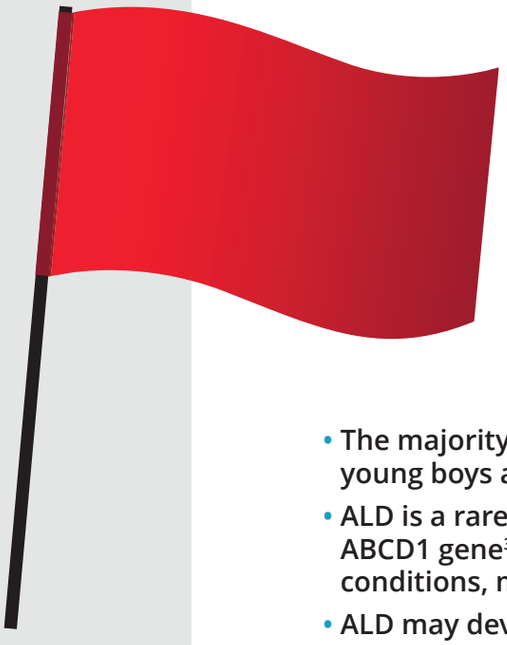

PRIMARY ADRENAL INSUFFICIENCY IN YOUNG BOYS IS A RED FLAG FOR ALD

- The majority of cases of antibody-negative primary adrenal insufficiency in young boys are caused by adrenoleukodystrophy (ALD)[1-3]
- ALD is a rare X-linked recessive disorder resulting from mutations in the ABCD1 gene[3]. The early symptoms of ALD are similar to other medical conditions, making diagnosis challenging[4]
- ALD may develop into cerebral ALD (CALD), a severe form of the disease[4,5]
- If untreated, CALD often leads to severe neurodegeneration and sometimes death within 2 to 3 years[5]

EARLY DIAGNOSIS OF ALD COULD SAVE A LIFE

- If CALD is caught early, lifesaving treatment is possible[6]
- ***Early detection is critical***

CHECK VERY LONG CHAIN FATTY ACID LEVELS (VLCFA)

- A simple VLCFA blood test can diagnose ALD[4]
- ***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[8]
- Occurs in about 1 in 20,000 males[3]
- Causes impaired peroxisomal β-oxidation of VLCFA, resulting in accumulation of VLCFA in plasma and all tissues[3]
- Early symptoms are variable and non-specific but may include behavioral or learning issues and audiovisual impairment[3,4]
- 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 peripheral nervous system in adults, and/or the rapidly progressive cerebral form (CALD)[1,7]

ABOUT 35-40% OF ALD CASES WILL CONVERT TO CALD[4]

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

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[9]

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

Codes and information obtained from Quest Diagnostics in the US. Please apply different country- and/or laboratory-specific information as appropriate.

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*

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® [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**. Bezman L et al. *Ann Neurol*. 2001;49(4):512-517. **8.** Mahmood A, Dubey P, Moser HW, Moser A. X-linked adrenoleukodystrophy: therapeutic approaches to distinct phenotypes. *Pediatr Transplant.* 2005;9(Suppl 7):55-62. **9.** Brett EM, Auchus RJ. Genetic forms of adrenal insufficiency. *Endocr Pract*. 2015;21(4):395-399.

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