Very Long-Chain and Branched-Chain Fatty Acids Profile

X-linked adrenoleukodystrophy (X-ALD) is a rare X-linked metabolic disorder caused by variants in the ABCD1 gene that cause a deficiency in adrenoleukodystrophy protein (ALDP) and subsequent accumulation of very long-chain fatty acids (VLCFAs). VLCFA accumulation occurs in plasma and all tissue types, but primarily affects the adrenal cortex and white matter of the brain and spinal cord, resulting in a range of clinical outcomes.

Adrenal insufficiency may be the initial presentation of X-ALD, and 21-hydroxylase antibody testing may confirm or exclude an autoimmune etiology. In X-ALD, 21-hydroxylase antibody testing results will be normal; therefore, males with adrenal insufficiency and normal 21-hydroxylase antibody testing should be tested for X-ALD (VLCFA profile).

For additional information on the testing strategy for X-ALD, refer to the ARUP Consult X-Linked Adrenoleukodystrophy topic.

Disease Overview

Incidence

1/14,700 live births

Penetrance

Neurologic symptoms are present in nearly 100% of males with the disease by adulthood.

Inheritance

X-linked

Test Interpretation

Biochemical testing (VLCFAs and BCFAs)

- Males: Elevated VLCFAs
- Females: Elevated VLCFAs in approximately 85% of heterozygous carriers

References


Related Information

X-Linked Adrenoleukodystrophy