

X-Linked Adrenoleukodystrophy on a Child

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Clinical Image

X-linked Adrenoleukodystrophy (X-ALD) is an inherited neurodegenerative disease due to the accumulation of Very Long-Chain Fatty Acids (VLCFA) in the cerebral White Matter (WM) resulting in severe inflammatory demyelination.

Clinical presentation depends on the phenotype that varies according to the patient's age.

MRI is the key imaging modality to evaluate ALD as it shows early abnormalities that precede clinical findings. It demonstrates symmetric white matter hyper intensity on T2-weighted and FLAIR images (Figures 1 and 2) and hypo- or isointensity on T1-weighted images. Post-contrast enhancement at the edges of affected white matter is predictive of disease progression. MRI presentation varies based on the involved regions and patterns of progression

Hematopoietic cell transplantation is indicated in the early stages of the disease. Corticosteroid replacement therapy is used in patients with adrenocortical insufficiency.

The images bellow were obtained in a 12 years old child, who presented with progressive motor and cognitive impairment.

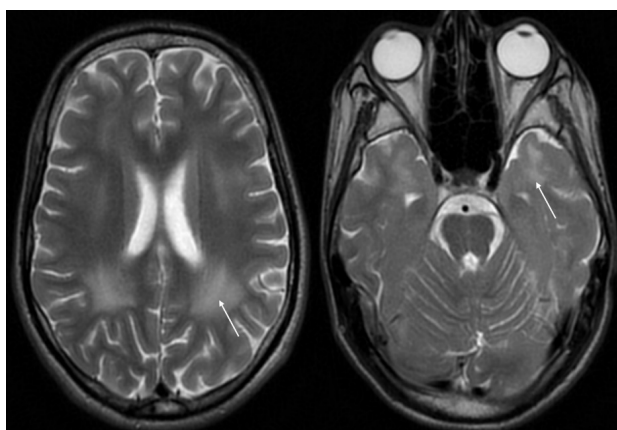


Figure 1: Axial T2-W cerebral MRI images demonstrating symmetric white matter hyperintensity in parietal and temporal lobes (arrows).

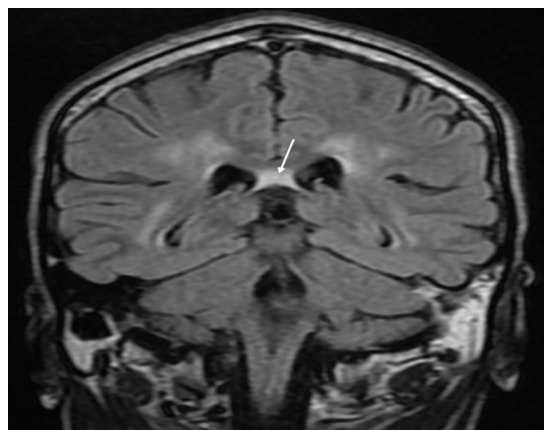


Figure 2: Coronal T2-W FLAIR cerebral MRI image demonstrating corpus callosum hyperintensity (arrow).