Teaching NeuroImages: MRI findings in X-linked adrenoleukodystrophy

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Figure MRI brain with spectroscopy at the time of diagnosis

(A) Fluid-attenuated inversion recovery (FLAIR) image shows symmetric confluent hyperintense white matter lesion in posterior cerebral hemispheres. (B) Postcontrast T1-weighted image shows symmetric linear enhancement, known as intermediate inflammatory zone, which is much less hyperintense on FLAIR and apparent diffusion coefficient map (C). (D) Magnetic resonance spectroscopy shows decreased N-acetylaspartate (NAA) and elevated choline (Cho) and lactate peaks. Cr = creatine.

A 6-year-old boy presented with 4 months progressive verbal comprehension difficulties, declining school performance, strabismus, and visual impairment. General examination noted moderate hypotension and hyper-pigmented skin. Pertinent neurologic examination abnormalities included difficulty with comprehension, decreased visual acuity (20/50 bilaterally), disconjugate gaze, bilateral dysmetria, and unsteady gait. CSF protein

was elevated (1.82 g/L). Brain MRI showed posterior cerebrum and brainstem abnormalities consistent with adrenoleukodystrophy¹ (figure). Morning cortisol was decreased (21 nM) and adrenocorticotrophic hormone stimulation test confirmed adrenal insufficiency. Diagnosis was confirmed by very long chain fatty acid analysis (elevated C26:0/C22:0 and C24:0/C22:0 ratios) and identification of pathogenic *ABCD1* mutation.

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AUTHOR CONTRIBUTIONS

Kenneth A. Myers: drafted the manuscript, prepared the figures, and approved the final manuscript as submitted. Jong M. Rho: reviewed and revised the manuscript and approved the final manuscript as submitted. Dr. Rho ensured that the patient history was accurately characterized. Xing-Chang Wei: reviewed and revised the manuscript and approved the final manuscript as submitted. Dr. Wei ensured that the patient's MRI findings were accurately characterized.

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