MRI in X-linked adrenoleukodystrophy

A 45-year-old man with no medical history presented with behavioral changes. He was withdrawn from his family and friends. Home and finances had fallen into disorder. He was disheveled and incontinent of urine and feces. He lacked insight into his cognitive decline. His skin was bronzed and the examination was notable only for upgoing plantar responses. Peroxisomal fatty acid profile was elevated at C24:0 at 138.9 nmol/mL (normal <91/4), elevated C26:0 at 5.05 (normal <1.39), and elevated C26:0/C22:0 ratio at 0.062 (normal <0.023). Images show abnormal high signal on fluid-attenuated inversion recovery (figure 1) with enhancement (figure 2).

X-linked adrenoleukodystrophy is a metabolic disorder characterized by the accumulation of very long-chain fatty acids in brain white matter. The disease is caused by a deficiency of peroxisomal very long-chain fatty acid oxidase, which results in the accumulation of very long-chain fatty acids. These fatty acids are then deposited in the brain, leading to neurocognitive impairment, behavioral changes, and ultimately death. The diagnosis is typically made through peroxisomal fatty acid profile and MRI imaging.

Saima Siddiqui, MD, Gauri Pawar, MD, Jeffery P. Hogg, MD
From West Virginia University, Morgantown.

Author contributions: Saima Siddiqui: original draft of paper, revision of paper after review of other coauthors, data acquisition, review of images. Jeffery P. Hogg: chose images used, wrote legends to images, helped with revision of original draft. Gauri Pawar: helped with original revision of paper.

Study funding: No targeted funding reported.

Disclosure: The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

Correspondence to Dr. Siddiqui: siddiqui@upmc.edu
MRI in X-linked adrenoleukodystrophy
Saima Siddiqui, Gauri Pawar and Jeffery P. Hogg
Neurology 2015;84;211
DOI 10.1212/WNL.0000000000001122

This information is current as of January 12, 2015

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/84/2/211.full

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
Leukodystrophies
http://n.neurology.org/cgi/collection/leukodystrophies
MRI
http://n.neurology.org/cgi/collection/mri

Permissions & Licensing
Information about reproducing this article in parts (figures,tables) or in its entirety can be found online at:
http://www.neurology.org/about/about_the_journal#permissions

Reprints
Information about ordering reprints can be found online:
http://n.neurology.org/subscribers/advertise

Neurology © is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2015 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.