Imaging findings in encephalocraniocutaneous lipomatosis

Figure Head CT and MRI

CT (A–C) and MRI (T2–fluid-attenuated inversion recovery [D, E], T1 [F]) show dilated left lateral ventricle and calcified cortex (B, E, arrows). Large heterogeneous epidural mass (asterisks; stable, likely fatty or proteinaceous material) has calcified margin continuous with calvarium (A, D, arrows). Lipomas (arrowheads) appear along the interhemispheric fissure (A) and cerebellopontine angle (C, E, F).

A man in his 30s with encephalocraniocutaneous lipomatosis (ECCL), hydrocephalus, ventriculoperitoneal shunt, and Lennox-Gastaut syndrome was seen for epilepsy. He had multiple facial subcutaneous nodules (lipomas), near-blindness bilaterally, and right spastic hemiparesis. He was fluent, dysarthric, and followed one-step commands. A partially thrombosed internal carotid aneurysm was found on imaging (not shown).

ECCL is a neurocutaneous syndrome resulting from ectomesodermal dysgenesis, characterized by choristomas (ocular tumors), hairless scalp lesions (nevus psiloliparus), lipomas (facial, intracranial, particularly at the cerebellopontine angle, or intraspinal), and calcifications (figure).¹ Half of patients have seizures, one-third have moderate mental retardation, and some have intracranial vascular malformations.²

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Author contributions: A. Svoronos was the primary author of the manuscript. Dr. Hirsch was the treating physician of the patient, developed the study concept, and revised the manuscript. Dr. Khandji performed the analysis and interpretation of radiologic images and revised the manuscript.

Disclosure: A. Svoronos reports no disclosures. Dr. Hirsch serves/has served as a consultant for Lundbeck Inc., Upsher-Smith, and Ikano Therapeutics Inc.; has received speaker honoraria from GlaxoSmithKline, Pfizer Inc, Lundbeck Inc., and UCB; serves on the editorial board of the *Journal of Clinical Neurophysiology* and as contributing editor for *Epilepsy Currents*; receives publishing royalties from *UpToDate* and for *Atlas of EEG in Critical Care* (Wiley-Blackwell, 2010); has served on speakers' bureaus for GlaxoSmithKline, UCB, Pfizer Inc, and Lundbeck Inc.; and receives/has received research support from Eisai Inc., Pfizer Inc, Lundbeck Inc., UCB, Upsher-Smith, the American Epilepsy Society, and the Epilepsy Foundation. Dr. Khandji reports no disclosures.

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Neurology 2011;77;694
DOI 10.1212/WNL.0b013e3182299fa9

This information is current as of August 15, 2011

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