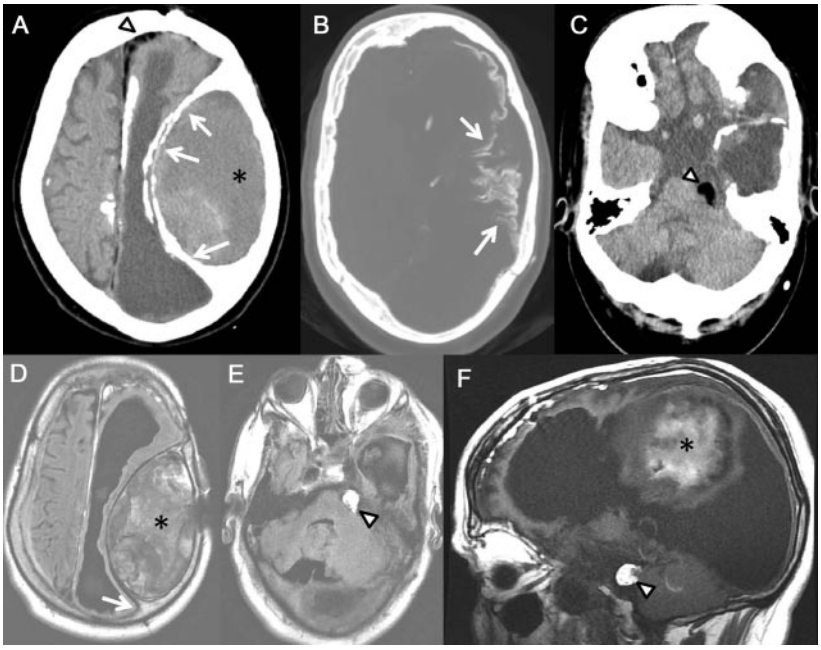
Imaging findings in encephalocraniocutaneous lipomatosis

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, 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.

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2. Moog U. Encephalocraniocutaneous lipomatosis. J Med Genet 2009;46:721–729.