Images in Clinical Practice

Encephalocraniocutaneous Lipomatosis

A 7-month female child presented with abnormal look. On examination child had cranial asymmetry, alopecia, nevus sebaceous over left scalp, epidermal verrucous nevus on neck, cafe-au-lait spots, multiple pigmentary patches over abdomen, bilateral corneal hazziness, skin tags at outer canthus and dermoid of both eyes since birth (*Fig. 1*). Child's weight was 5.8 kg., length 64 cm, upper segment 39 cm, head circumference 43.5 cm with normal developmental age. Routine hematology, B-Scan of eyes, ECG, ECHO and abdominal USG were normal. CT scan head revealed-lipoma (HU value –40 to –60) at left cerebello pontine angle, 10 mm × 7 mm × 5 mm size, with dilated left ventricle, cortical atrophy (*Fig. 2*). A diagnosis of encephalocraniocutaneous lipomatosis was made.

Encephalocraniocutaneous lipomatosis is a rare congenital neurocutaneous disease. It is characterized by unilateral lipomatous hamartoma of scalp, eyelid and outer globe of

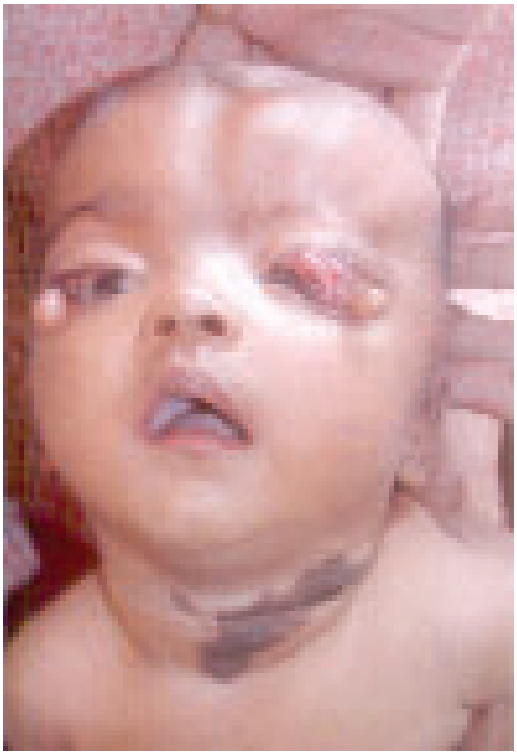

Fig. 1. Clinical Photograph showing the features of encephalocraniocutaneous lipomatosis.

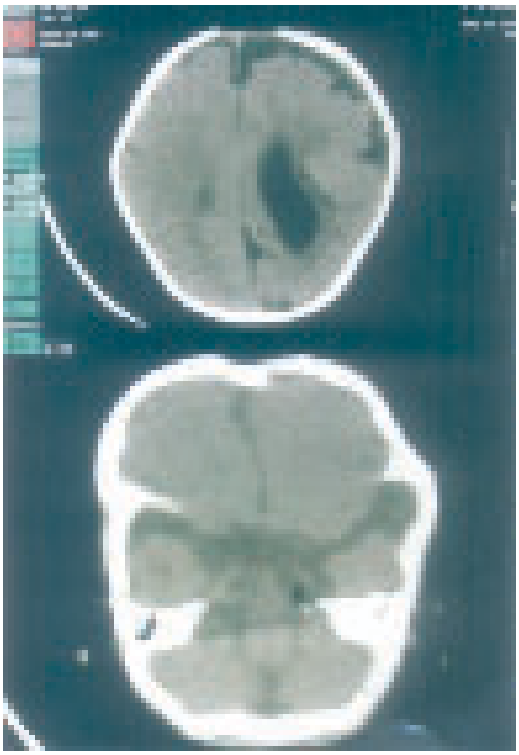

Fig.2. CT scan depicts dilated left lateral ventricle with lipoma (arrowhead) at left cerebello-pontine angle.

eye, ipsilateral porencephalic cyst, intracranial lipoma, cortical atrophy, cranial asymmetry, developmental delay and mental retardation. The clinical picture may vary from patient to patient. Mental status varies from totally normal to severe mental retardationr seizure may be associated in some cases. Bilateral cutaneous and visceral involvement is also rarely reported. The pathogenesis remains unknown. Dysgenesia of the cephalic neural crest and the anterior neural tube is a most widely accepted theory.

Rajiv Rathoriya,
Jyotsna Shrivastava,
Department of Pediatrics,
Gandhi Medical College,
Bhopal, Madhya Pradesh, India.
E-mail:shrivastav_rajendra@rediffmail.com