**RELATO DE CASO**

# Encephalocraniocutaneous Lipomatosis: Report on a Brazilian girl

## *Lipomatose Encéfalo-Crânio-Cutânea: Relato de caso de uma garota brasileira*

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**Abstract** Encephalocraniocutaneous lipomatosis (ECCL) is a rare congenital disorder which belongs to the group of neurocutaneous syndromes. This condition comprises seizures beginning in infancy, mental retardation, and unilateral cutaneous and ophthalmologic lesions with ipsilateral cerebral manifestations. It is not clear if ECCL is a localized form of the Proteus syndrome or not, and differential diagnoses include other neurocutaneous disorders, such as oculocerebrocutaneous syndrome (OCC), neurofibromatosis type I (NF I) and epidermal naevus. We reported a case of a Brazilian girl with clinical signs compatible with ECCL.

**Keywords** Encephalocraniocutaneous Lipomatosis (ECCL); Fishman syndrome; Brain anomalies.

**Resumo** Lipomatose encéfalo-crânio-cutânea (LECC) é uma doença congênita rara do grupo de síndromes neurocutâneas. Esta condição tem como principais características: convulsões na infância, atraso mental, lesão cutânea unilateral e lesão oftalmológica com manifestação cerebral ipsilateral. Não está claro se LECC é uma forma localizada da síndrome de Proteus, ou não, e diagnóstico diferencial inclui outras doenças neurocutâneas, tais como: síndrome oculocerebrocutânea (OCC), neurofibromatose tipo I (NF I) e nevus epidérmico. Nós relatamos o caso de uma menina brasileira, com sinais clínicos compatíveis com LECC.

**Palavras-chave** Lipomatose encéfalo-crânio-cutânea, LECC, síndrome Fishman, anomalias cerebrais

**Introduction**

Encephalocraniocutaneous lipomatosis (ECCL) is a rare disorder of unknown etiology, first described by Haberland and Perou in 1970[1]. The main features of this condition are macrocephaly, alopecia, lipodermoids involving the conjunctiva, sclera or eyelids, and lipomatous swellings over the cranium or face. Mental retardation is a common feature and a CT brain scan shows cerebral atrophy, sometimes unilateral, or porencephalic cysts. Around 34 cases have been described in the literature; the neurocutaneous findings being ipsilateral in most cases[2,3,4].

We have reported a case of a patient with features according to the diagnosis of ECCL.

**Case Report**

The girl (Fig. 1A-C) was the first child of a G1P1 19-year old woman and her 26- year old nonconsanguineous husband. Pregnancy was complicated by vaginal bleeding in the fourth and fifth months. Delivery was by means of cesarean section. Birth weight was 3,530 g (50th-75th centile), length 50.5 cm (50th centile), and head circumference 36 cm (98th centile). There were no peri or neonatal complications.

Clinical examination at age 3 6/12 years showed a weight of 16.100 Kg (75th centile), height of 98 cm (50th-75th centile), and OFC of 50 cm (50th centile). She had a scalp defect at the right, prominent forehead, and asymmetric face. The right side presented a soft tissue mass in the parietal area, tags in the malar area and upper palpebral fissures, and asymmetric chest with a hypercromic skin area. Her neuropsychomotor development was normal. Conjunctival angiomatosis and abnormal iris and cornea in the right eye and lipodermoids affecting the upper outer quadrant of the left eye were observed. Seizures have being treated with anticonvulsants. A magnetic resonance image scan and computerized tomography scan (Fig. 2 A-C) revealed skull hamartoma, arachnoidal cyst, intracranial lipomas, cerebral hemiatrophy, and venocapilar hemangioma in the right temporo-occipital region. Skin biopsy did not show any histological alterations of haemangioma or naevus. Abdominal ultrasound, intravenous pyelography, echocardiogram, and G-banded karyo-

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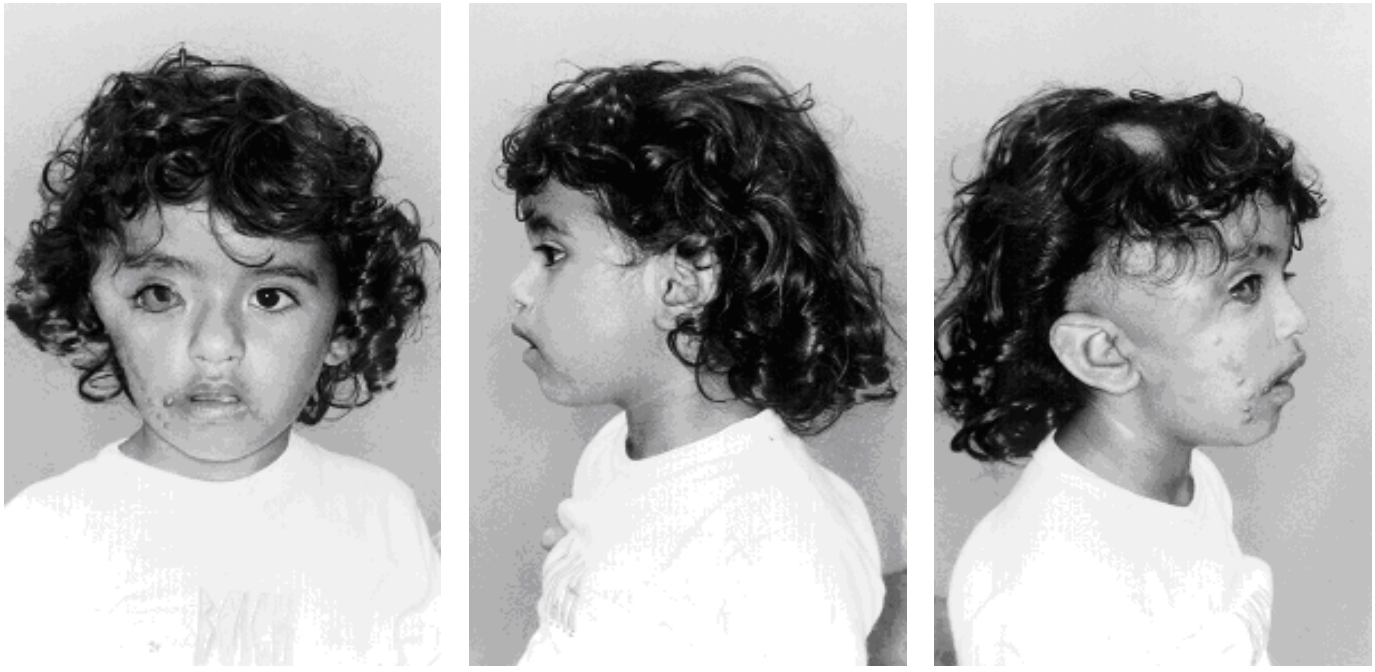

**Fig. 1A-C:** Clinical aspects of the patient

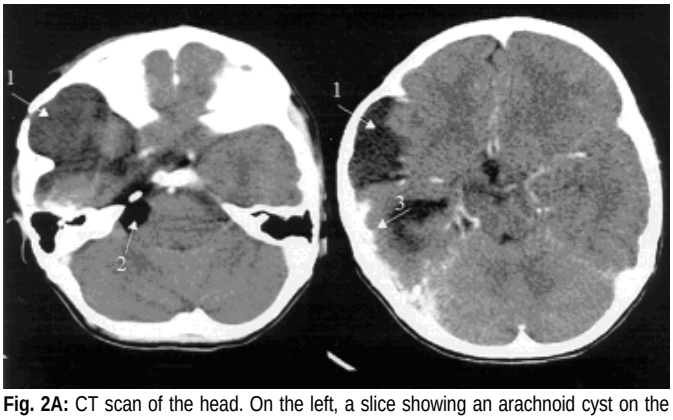


**Fig. 2A:** CT scan of the head. On the left, a slice showing an arachnoid cyst on the temporal fossa, expanding the cranial vault (arrow 1), associated with temporal lobe atrophy. Adjacent to the petrous bone there is a lipoma (arrow 2). On the right a slice showing the superior portion of the arachnoid cyst and cortical enhancement with vascular shape on the posterior temporal lobe (arrow 3).

type were normal.

**Discussion and Conclusion**

The patient clinically studied in this report, closely resembles that described by Haberland and Perou[1] and Fishman et al.[5] . The similarities included unilateral cutaneous cranial and ipsilateral facial lesions consisting of papular lesions of the face, lipodermoids of the eye, brain abnormalities, and scalp lesion.

The clinical features of ECCL (OMIM[6] 176920) overlap with other neurocutaneous syndromes and the main differential diagnosis includes oculocerebrocutaneous syndrome (OCC - OMIM[6] 164180), neurofibromatosis type I (OMIM[6] 162200), epidermal naevus syndrome (OMIM[6] 163200), and Proteus syndrome (OMIM[6] 176920) (Table 1). In OCC syndrome, the presence of ano/microphthalmia, skin defects and skin tags, as well as no facial lipomas and scalp alopecia, are helpful for the diagnosis. In this syndrome, CNS malformations are limited to intracranial cysts and agenesis of the corpus callosum[7]. Although our patient presented intracranial cysts, other CNS abnormalities were found, which are uncommon signs in OCC syndrome. Neurofibromatosis type I is a common autosomal dominant condition where cafe au lait patches, peripheral neurofibromata, absent in our patient, are the most common features. Epidermal naevus syndrome presents as main features lesions of midline of the face associated with seizures and mental retardation. In our case, the facial midline was preserved. The Proteus syndrome is an

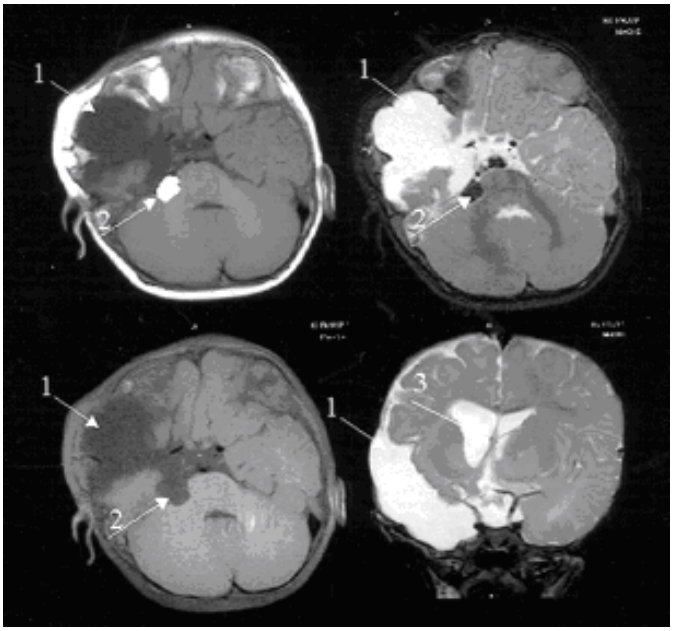


**Figure 2B:** Magnetic Resonance Image of the head. On the upper left a T1-weighted image, on the upper right a T2-weigheted image, on the lower left a T1-weighetd image with fat saturation. All the three axial slices are on the same level. On the lower right a coronal T2-weighetd slice is shown, through the temporal lobe. Note the arachnoid cyst on the temporal fossa, T1 hypointense and T2 hyperintense (arrows 1). The lipoma is seen next to the pons (arrows 2), bright on T1-w, black on T2-w and excluded on the fat saturation image. The coronal slice shows better the relationship between the arachnoid cyst and temporal lobe atrophy with ventricle enlargement (arrows 3).

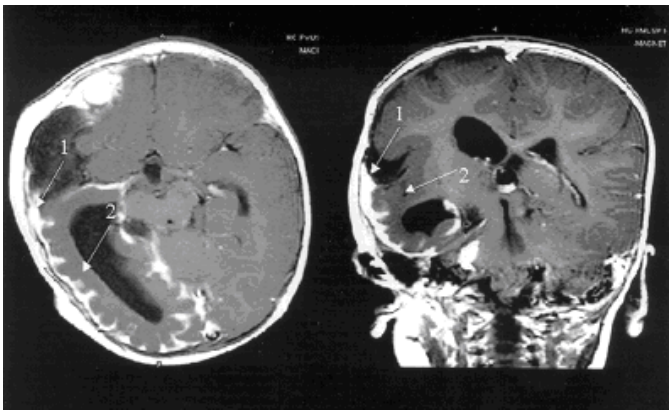


**Figure 2C:** Magnetic Resonance Image, with gadolinium injection (i.v. 0.2 ml/kg). T1-weighted images, with an axial slice on the left and a coronal slice on the right. A cortical vascular enhancement is seen on the temporal lobe (arrow 1) what suggests veno-capilllary hemangioma over a thick dysplastic cortex (arrow 2).

**Table 1:** Comparison of the clinical signs in OCC syndrome, NF I, Epidermal naevus syndrome, Proteus syndrome, and ECCL syndrome with those in the present patient

| Clinical Signs | OCC | NF I | Epidermal naevus | Proteus | ECCL | Our patient |
|---|---|---|---|---|---|---|
| Macrocephaly | - | + | + | + | + | - |
| Facial asymmetry | + | + | + | + | + | + |
| Skull hyperostoses | - | - | - | + | - | - |
| Focal alopecia | + | - | - | - | + | + |
| Orbital cyst | + | - | - | - | - | + |
| An/microphtalmia | + | - | - | - | + | - |
| Cloudy cornea/choristoma | + | - | + | + | + | + |
| Eyelid coloboma | + | - | + | - | + | - |
| Lisch nodules | - | + | - | - | - | - |
| Cranial hamartoma/lipoma | + | - | - | - | + | + |
| Facial skin tags | + | + | - | - | + | + |
| Neurofibromas | - | + | - | + | + | - |
| Café au lait patches | - | + | + | + | - | - |
| Rib defects | + | - | - | - | - | - |
| Asymmetry of limbs | - | + | + | + | - | - |
| Partial gigantism of hands and/or feet | - | - | - | + | - | - |
| Vascular anomalies | - | + | + | + | - | - |
| Mental retardation | + | + | + | + | + | - |
| Seizures/EEG anomalies | + | + | + | + | + | + |
| Abnormal cerebral ventricles | + | + | + | + | + | - |
| Intracranial cysts | + | + | - | - | + | + |
| Callosal a/hypoplasia | + | + | - | + | - | - |
| Cerebral calcifications | - | - | + | - | + | - |

OCC=oculocerebrocutaneous syndrome; NF I=neurofibromatosis type I; ECCL=encephalocraniocutaneous lipomatosis

overgrowth syndrome in which every feature of ECCL can be found. Brain alterations are rare[8]. While ECCL is non-progressive, unilateral, and limited to the head, the Proteus syndrome is progressive, bilateral, asymmetric, and involves the head, trunk, and limbs[9]. Some authors suggest that these two syndromes may represent variability of the same genetic entity, and that ECCL may be a more localized form of the Proteus syndrome[10]. These same authors have reported chromosomal mosaicism, or a somatic mutation lethal in nonmosaic states. On the other hand, McCall et al.[9] have maintained them as separate entities.

Although the presence of haemangioma in our patient is a typical sign of the Proteus syndrome, other main features showed in this condition such as asymmetry of limbs, partial gigantism of hands and/or feet and bone hypertrophy were not observed in our propositus. We think that the constellation of abnormalities observed in our patient represents the ECCL syndrome and, we believe that this is a distinct entity from the Proteus syndrome.

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