

Case report

Encephalocraniocutaneous lipomatosis (Fishman syndrome): A rare neurocutaneous syndrome

Mohammad Sharifi*, Maral Namdari

Eye Research Center, Mashhad University of Medical Sciences, Mashhad, Iran

Received 11 May 2016; accepted 12 June 2016

Available online 15 July 2016

Abstract

Purpose: To report a rare case of encephalocraniocutaneous lipomatosis (ECCL) presented with characteristic multiple organ involvement.

Methods: A 7-day-old white Iranian girl was referred with ocular, skin and brain abnormalities.

Results: The findings of nevus psiloliparus, eyelid choristoma and intracranial lipoma were consistent with ECCL.

Conclusion: Since the skin and ocular manifestations can be easily observed at birth examination, pediatricians and ophthalmologists should be aware of this condition.

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Keywords: Choristoma; Encephalocraniocutaneous lipomatosis; Neurocutaneous; Nevus psiloliparus

Introduction

Encephalocraniocutaneous lipomatosis (ECCL), also known as Fishman syndrome, is a rare congenital neurocutaneous disease that commonly involves ectomesodermal tissues, such as eye, skin, and central nervous system.^{1,2} Nevus psiloliparus (NP), a rare skin anomaly characterized by alopecia and an excessive amount of fat tissue is the hallmark of ECCL.² Eyelid choristoma is the predominant ocular feature, while intracranial lipoma is the major central nervous system (CNS) manifestation. We present a 7-day-old girl with ocular, skin, and CNS malformations consistent with ECCL.

Case report

A 7-day-old white Iranian girl was referred to the ophthalmology clinic for ophthalmic evaluation. She was a full-term

infant from healthy, non-consanguineous parents after an uncomplicated pregnancy. She was born with cesarean-section (C/S) due to previous C/S. Family history was negative. General physical examination showed a length of 49 cm, weight of 3.2 kg and head circumference of 35 cm, which were in normal ranges. Routine laboratory examinations in pregnancy were normal. Maternal screening tests for trisomy 13, trisomy 18, and Down Syndrome were negative. Ophthalmic examination showed multiple skin-colored papules and pedunculated lesions located on her right and left eyelids, and lateral canthus, right upper eyelid coloboma, right bulbar conjunctival hypertrophy, right limbal dermoid, corneal haziness and corneal peripheral vascularization extending from 2 to 6 o'clock in the left eye (Fig. 1).

Fundus examination of the right eye was not possible due to corneal opacity, but left fundus showed a peripapillary hypopigmented creamy-white irregular choroidal lesion (6–7 disc diameters). Optic disc and peripheral retina were normal (Fig. 2). Ultrasonographic scan showed high intensity echo spikes and highly reflective choroidal mass with posterior acoustic shadowing in favor of posterior globe calcification (Fig. 3).

* Corresponding author.

E-mail address: sharifim@mums.ac.ir (M. Sharifi).

Peer review under responsibility of the Iranian Society of Ophthalmology.



Fig. 1. Ocular and dermal findings include: right upper lid coloboma, multiple soft skin-colored pedunculated periocular lesions, bulbar conjunctiva hypertrophy, corneal clouding and right limbal dermoid and soft, elevated, area of patchy hair loss in the frontotemporal region (nevus psiloliparus).

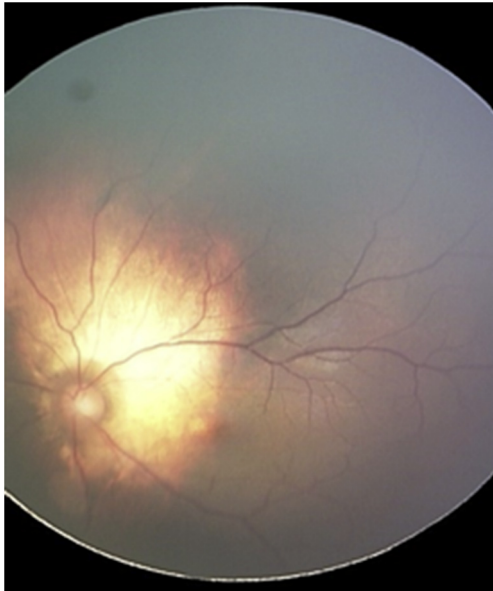


Fig. 2. Left fundus Retcam showing peripapillary hypopigmented creamy-white irregular choroidal lesion with normal optic disc and peripheral retina.

Dermatological examination showed a soft, elevated area of patchy hair loss on the right scalp extending to the forehead without signs of inflammation or scarring, clinically compatible with NP (Fig. 1).

Routine laboratory examination including complete blood count, erythrocyte sedimentation rate, and C-reactive protein were normal. TORCH screening serology tests were negative. Axial non-contrast computed tomography (CT) scanning of the brain and orbit revealed fat density lesions in subcutaneous tissues of the right temporal region and ipsilateral suprasellar, and cerebellopontine angle hypodense lesion suggestive of lipomas. Focal calcifications were seen in the posterior globe of both eyes as well as extraconal area of the right eye. Eyeballs seemed normal in size (Fig. 4).

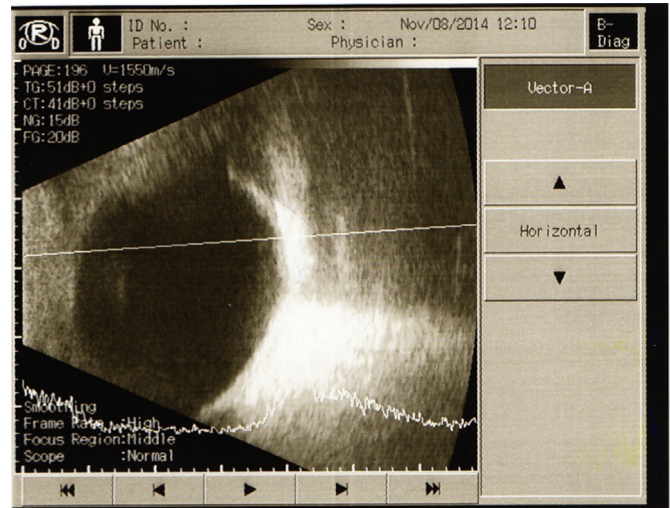


Fig. 3. Ultrasound of both eyes showed A scan with high intensity echo spikes and highly reflective choroidal mass with posterior acoustic shadowing in favor of posterior globe calcification.

Echocardiography revealed a patent foramen ovale. The constellation of these clinical and imaging findings led to a diagnosis of ECCL.

Discussion

The ECCL or Fishman syndrome is presented with congenital skin, eye and brain lesions. The etiology is most likely due to dysgenesis of the anterior neural tube and cephalic neural crest.³ All reported cases of ECCL are sporadic. A nonhereditary, autosomal mutation that may survive only in a mosaic state may be a cause of the clinical picture of ECCL.⁴ The first description of this syndrome as ‘Encephalocraniocutaneous lipomatosis’ was by Haberland and Perou in 1970.⁵ Afterwards, Fishman *et al*⁶ in 1987 reported more cases of ECCL. In a literature search on PubMed database, 77 patients with Fishman syndrome were found (accessed on September 2013). There is no clear gender, racial, or geographical predilection. The diagnosis of ECCL is mainly based on history, clinical examination, and imaging studies. MacLaren *et al* in 1995 and Hunter in 2006⁷ have laid down diagnostic criteria, but Moog in 2009 proposed revised diagnostic criteria for ECCL.⁸ Table 1 shows Moog's revised criteria for diagnosis of ECCL.

Ocular lesions are always present and consist of conjunctival choristoma, with or without associated anterior chamber anomalies.⁷ Persistent hyaloid vessels, lens dislocation, iris dysplasia, aniridia, colobomas, microphthalmia, ocular calcifications, and optic disc pallor have also been reported.^{9,10} The most characteristic scalp lesion in ECCL is NP, a soft, subcutaneous mass with demarcated area of alopecia. Dermal lesions in the face are multiple papular or polypoid nodules. Histologically, facial and scalp lesions are hamartomas and choristomas. These lesions are unilateral in most cases, but they can be bilateral.⁸ Brain abnormalities include cerebral and spinal lipomas, intracranial calcifications, abnormal

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