ARCHIVOS DE LA SOCIEDAD ESPAÑOLA DE OFTALMOLOGÍA

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Short communication

Haberland syndrome with bilateral ocular involvement☆

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ARTICLE INFO

Article history:
Received 30 March 2020
Accepted 8 July 2020
Available online 6 November 2020

Keywords:
Encephalocraniocutaneous lipomatosis
Haberland syndrome
Choristoma
Arachnoid cyst

ABSTRACT

Haberland syndrome or encephalocutaneous lipomatosis is a very uncommon syndrome that is characterised by changes in the skin, eye, and central nervous system. It was first described in 1970 by Haberland and Perou, with about 60 cases having been reported since then. A case is reported of a 14-week-old male diagnosed with Haberland syndrome with bilateral ocular involvement in the form of palpebral coloboma and choristomas.

Síndrome de Haberland con afectación ocular bilateral

Palabras clave:
Lipomatosis encefalocraneocutánea
Síndrome de haberland
Coristoma
Quiste aracnoideo

RESUMEN

El síndrome de Haberland o lipomatosis encefalocraneocutánea es un síndrome muy infrecuente caracterizado por la triada clásica de afectación cutánea, ocular y del sistema nervioso central. Fue descrito por primera vez en 1970 por Haberland y Perou,[1] habiéndose reportado unos 60 casos desde entonces.[2] Presentamos un caso de un varón de 14 semanas diagnosticado de síndrome de Haberland con afectación bilateral ocular en forma de coloboma palpebral y coristomas.

☆ Please cite this article as: Zamorano Martín F, García Lorente M, Rocha de Lossada C, Rachwani Anil R, Santos Ortega A, Escudero Gómez J. Síndrome de Haberland con afectación ocular bilateral. Arch Soc Esp Oftalmol. 2021;96:45–47.

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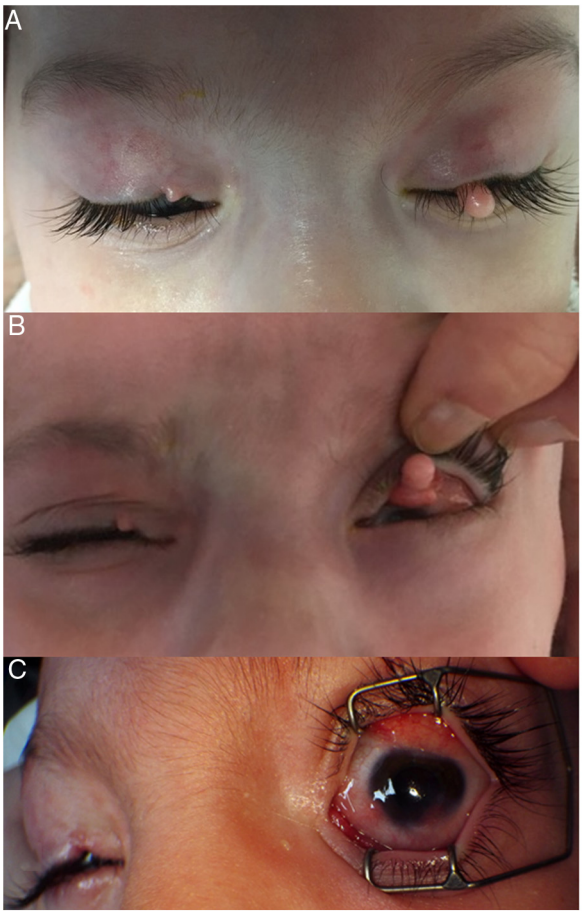

Fig. 1 – A) Palpebral coloboma in the inner third of the upper eyelid of both eyes. B) Pedunculated tumor dependent on the left upper tarsal palpebral conjunctiva. C) Soft pink lesions on lower bulbar conjunctiva of both eyes that invade the cornea without affecting the visual axis.

Introduction

The Haberland syndrome or encephalocranial lipomatosis is a very rare syndrome characterized by the classic triad of skin, eye (in the form of unilateral choristoma and palpebral coloboma) and central nervous system involvement. It was first described in 1970 by Haberland and Perou,[1] and since then about 60 cases have been reported.[2]

Case report

A 14-week male patient, first child of non-blood parents. Born at full term by Cesarean section. On examination, a 2 cm right temporal alopecia, palpebral coloboma in the inner third of the upper eyelid of both eyes (Fig. 1A), a pedunculated tumor dependent on the left upper palpebral tarsal conjunctiva (Fig. 1B) and soft pink lesions on the lower bulbar conjunctiva of both eyes were observed invading the cornea without compromising the visual axis (Fig. 1C). These lesions and the palpebral pedicle tumor were finally catalogued as complex choristomas by pathological anatomy.

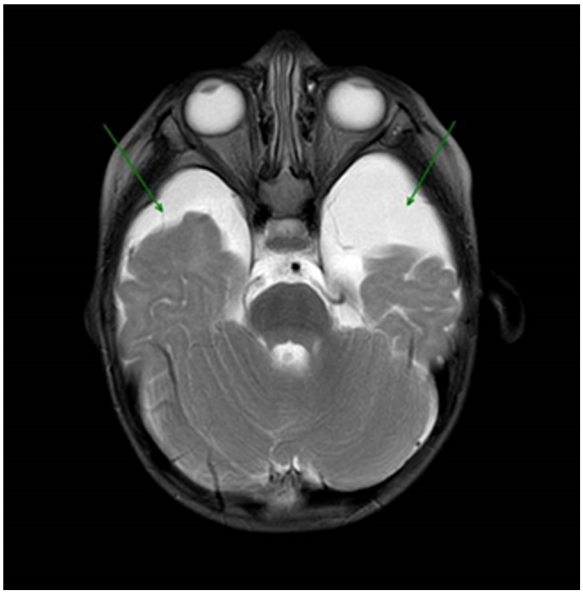

Fig. 2 – T2 axial brain MRI. Significant increase of bilateral temporal predominant subarachnoid spaces, compatible with bilateral temporal arachnoid cysts, more significant in the left side.

The patient exhibited good visual behaviour. The preferential gaze test produced a visual acuity of 8 cycles per degree in binocular vision and 4 cycles per degree with each eye in monocular vision. In fundus examination, a papilla with 0.4 excavation and lower depigmentation stood out.

In view of the suspected neurocutaneous picture, a cerebral magnetic resonance was requested in which an increase of the temporal subarachnoid spaces was appreciated bilaterally with arachnoid cysts (Fig. 2), being more significant in the left side, where it exhibited an approximate thickness of 3 cm. An increase in the parietal subarachnoid spaces was also observed bilaterally.

Serological tests for HIV and TORCH were negative. The rest of the examination did not provide new data of interest. The diagnosis of Haberland syndrome with bilateral ocular affectation was made.

Discussion

The Haberland syndrome or encephalocranial lipomatosis is a very rare syndrome characterized by the classic triad of skin, eye and central nervous system involvement. Its etiology is unknown, although it is postulated that it may be caused by a mutation in a gene during embryogenesis, being only compatible with life in the form of mosaicism.[3–7]

The most typical ocular involvement is in the form of unilateral and ipsolateral coristoma to skin lesions, which can cause amblyopia by inducing corneal opacities, refractive defects or both.[3,5] A less frequent form of presentation is palpebral coloboma1.

At the central nervous system, arachnoid cysts appear in the middle cranial fossa, dysplasia or cortical atrophy with adjacent ventricular dilation and cortical calcifications.[3,4,8]

60% of patients present epilepsy and 40% psychomotor retardation.[6]

An atypical case of encephalocraneocutaneous lipomatosis diagnosed at a very early age is presented. The case presents palpebral coloboma and bilateral choristoma, differing from most published cases in which these findings are exhibited unilaterally.[3,5] Associated clinic and imaging tests are essential for classifying this pathology.

In summary, since the Haberland syndrome or encephalocranial lipomatosis is a rare disease with great clinical heterogeneity, the ophthalmologist may be the first specialist consulted to assess a patient with this pathology. The presence of choristomas together with alopecia or other ipsolateral skin alteration should rule out a central nervous system condition by means of imaging tests.

Conflict of interest

None of the authors have any conflict of interest.

REFERENCES

1. Haberland C, Perou M. Encephalocraniocutaneuos lipomatosis. Arch Neurol. 1970;22:144–5.
2. Selçuk Ö, Ceyhun S, Cumhur K, Hanife G, Mustafa K, Ali Nail Dl, et al. Encephalocraniocutaneous Lipomatosis: Haberland Syndrome. Am J Case Rep. 2017;18:1271–5.
3. Rubegni P, Risulo M, Sbano P, Buonocore G, Perrone S, Fimiani M. Encephalocraniocutaneous lipomatosis (Haberland syndrome) with bilateral cutaneous and visceral involvement. Clin Exp Dermatol. 2003;28:387–90.
4. Brown KE, Goldstein SM, Douglas RS, Katowitz JA. Ecephalocraniocutaneous lipomatosis: A neurocutaneous syndrome. J AAPOS. 2003;7:148–9.
5. Amor DJ, Kornberg AJ, Smith LJ. Ecephalocraniocutaneous lipomatosis (Fishman syndrome): A rare neurocutaneous syndrome. J Paediatr Child H. 2000;36:603–5.
6. Donaire A, Carreno M, Bargalló N, Setoanín X, Agudo R, Martín G, et al. Presurgical evaluation and cognitive functional reorganization in Fishman síndrome. Epilepsy Behav. 2005:440–3.
7. Tambe KA, Ambekar SV, Bafna PN. Delleman (oculocerebrocutaneous) síndrome: Few variations in a classical case. Eur J Paediatr Neurol. 2003;7:77–80.
8. Nowaczyk MJ, Mernagh JR, Bourgeois JM, Thompson PJ, Jurriaans E. Antenatal and postnatal findings in encephalocraniocutaneous lipomatosis. Am J Med Genet. 2000;91:261–6.