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Sturge–Weber syndrome associated with naevus of Ota

SANTI MARIA RECUPERO,
SOLMAZ ABDOLRAHIMZADEH,
MARCO DE DOMINICIS,
ROBERTO MOLLO

Abstract

The association of Sturge–Weber syndrome with naevus of Ota is an infrequently reported phenomenon and there are only four previously described cases in the literature. In this paper we briefly review the literature regarding the coexistence of vascular and pigmentary naevi and present an additional patient with the association of the Sturge–Weber syndrome and naevus of Ota.

Key words Sturge–Weber syndrome, naevus of Ota

Case report

A 15-year-old girl was examined. Her parents were not blood relatives and there was no family history of neurocutaneous disease. The patient was the 3.8 kg product of a normal pregnancy and delivery. A port wine stain on the upper right portion of the face and the periorbital region was present at birth. The patient also presented a bluish discoloration of the right side of her face distributed over the lower eyelid, periocular region, temporal area and right ear (Fig. 1).

Magnetic resonance imaging revealed hyperplasia of the cavernous sinus due to vascular malformations in association with the facial angioma. On ophthalmological examination the patient's best corrected visual acuity was 20/25 with +1sf = + 0.5cil 110° OD and 20/20 OS with no correction. The anterior segment was normal in the left eye. The findings in the anterior segment of the right eye consisted of vast areas of bluish pigmentation of the conjunctiva and episclera, heavily pigmented iris that appeared thickened with obscured crypts and radial folds (Fig. 2) and poor pupillary response to pharmacological dilatation. Direct and consensual light reflexes were normal in both eyes. Intraocular pressure measured by applanation tonometry was 16 mmHg in both eyes.

Gonioscopy revealed an open angle on both sides but there was diffuse hyperpigmentation of the trabecular meshwork on the right (Fig. 3). Fundus examination of the right eye showed slight nasal peripapillary hyperpigmentation and a choroidal angioma of approximately one papillary diameter in the extreme periphery of the temporal quadrant. The left fundus was normal. Visual fields of both eyes were normal.

Discussion

The coexistence of cutaneous haemangioma and pigmentary naevi was termed phakomatosis pigmentovascularis in 1947.[1] These forms have been classified into four types and further divided into (a) localised (cutaneous) or (b) systemic on the basis of the presence or absence of systemic involvement[2] (Table 1). The patient described in our report can be classified as type IIb. To our knowledge only four similar cases have been previously described in the literature.[3–5] Some 20 other cases in the Japanese literature have been included in this group but not all patients presented melanosis bulbi or had the sole association of Sturge–Weber syndrome and naevus of Ota. Indeed, Klippel–Trenaunay syndrome and naevus anaemicus were present in numerous cases.

Recent reports consider that the embryopathogenesis of the skin, leptomeningeal, and choroid and brain lesions of Sturge-Weber syndrome can be explained by a malformation of the primordial vascular plexus.[6] In contrast, the hyperpigmentation of the skin, episclera and uvea in naevus of Ota is thought to be a result of maldevelopment and abnormality in the migration of neural crest cells, particularly melanocytes.[7] In considering phakomatosis pigmentovascularis, there is evidence supporting developmental abnormalities of vasmotor nerve cells and melanocytes that originate in the embryonal neural crest. Kitamura *et al.*[8] emphasised the electron microscopic differences between naevus flammeus in phakomatosis pigmentovascularis and port wine stain not accompanied by pigment abnormalities. Later, immunohistochemical studies showed the presence of perivascular nerves in port wine stains associated with phakomatosis pigmentovascularis, supporting this finding.[9] Therefore, it has been postulated that the association of pigmentary and vascular naevi is caused by functional disorders of vasomotor

S.M. Recupero ✉
S. Abdolrahimzadeh
M. De Dominicis
R. Mollo
Università di Roma 'La Sapienza'
II Clinica Oculistica
Policlinico Umberto I
Viale del Policlinico
I-00161 Rome
Italy
Tel: +39-6-490296
Fax: +39-6-4457706

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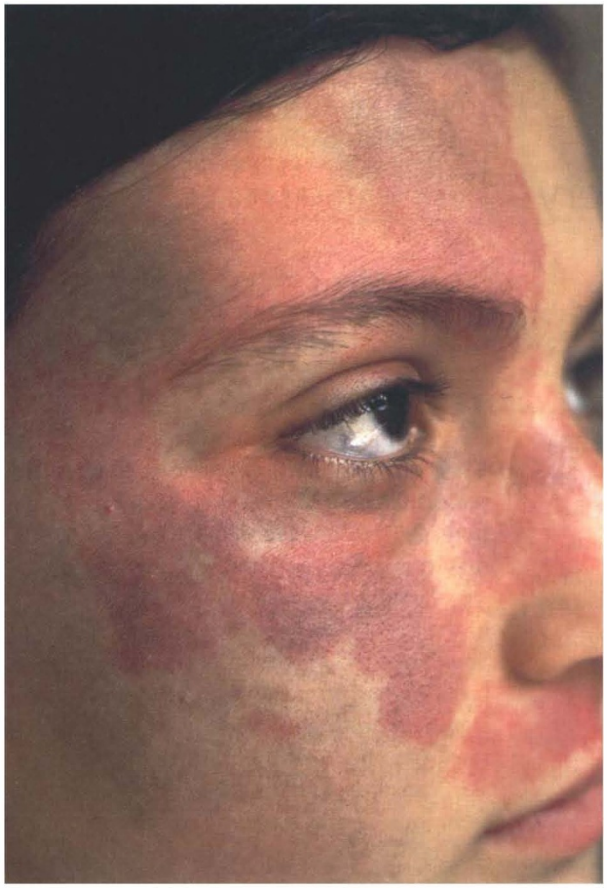

Fig. 1. *Port-wine stain and bluish discoloration of the right side of the face distributed over the lower eyelid, periocular region and temporal area.*

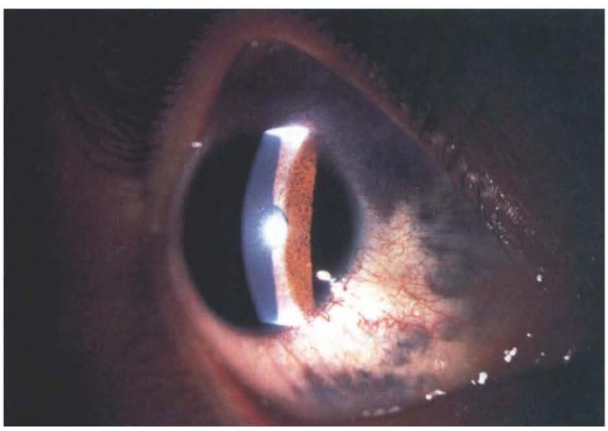

Fig. 2. *Right eye. There are vast areas of bluish pigmentation of the conjunctiva and episclera. The heavily pigmented iris appears thickened with obscured crypts and radial folds.*

Table 1. *Classification of phakomatosis pigmentovascularis*

Type	Features
I a, b[a]	Naevus flammeus and naevus pigmentosum et verrucosus
II a, b	Naevus flammeus, Mongolian spots,[b] ± naevus anaemicus
III a, b	Naevus flammeus, naevus spilus, ± naevus anaemicus
IV a, b	Naevus flammeus, Mongolian spots, naevus spilus, ± naevus anaemicus

From Hasegawa and Yasuhara.[2]
[a]a, cutaneous disease only; b, cutaneous and systemic disease.
[b]Mongolian spots (otherwise naevus of Ota or oculodermal melanocytosis).

Fig. 3. *Right eye. The angle is open but not wide, with diffuse hyperpigmentation of the trabecular meshwork.*

nerve cells and abnormal melanocytes, which originate in the embryonal neural crest. However, the pathogenetic mechanism becomes more complicated where there is systemic involvement (type b) as not all sites of origin can be attributed to the neural crest. Indeed, in Sturge–Weber syndrome there are lesions that have cells derived from the embryonal mesenchyme. Furthermore, the migration and differentiation of neural crest cells is influenced by the embryogenic environment and extracellular macromolecules such as glycosaminoglycans, fibronectin and collagen, rendering the pathogenetic mechanism even more complex.

In conclusion, the numerous cases of phakomatosis pigmentovascularis and the five cases of Sturge–Weber syndrome and naevus of Ota reported show that there is probably a common pathogenesis. However, further investigation into the molecular biology of neural crest development and angiogenesis of the central nervous system is warranted to determine the aetiology of these coexisting neuro-oculo-cutaneous disorders.

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