Case 1: A Child with Phakomatosis Pigmentovascularis

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CASE SUMMARY

History

A three-year old girl was referred to the dermatology clinic because of the presence of extensive vascular lesions on right side of face, trunk and limbs and extensive blue patches noted since birth.

She had history of generalized tonic clonic seizure since eight months of age, and has been put on anticonvulsant (sodium valproate). She had been seizure-free for the past one year. Investigations were done by neurosurgeons which showed the presence of extensive cerebral angioma on the right side. However, because of the extent of the lesion, interventions such as surgery or gamma-knife radiotherapy had been deemed unfeasible.

The child had also been assessed by the ophthalmologist and was found to be suffering from myopia and astigmatism. So far there was no evidence of glaucoma or buphthalmos. She also had problems with spatial relationship and fine motor control of her upper limbs. Training by the occupational therapists resulted in significant improvement.

She had been referred for child assessment, and was found to have normal developmental milestone for gross motor and speech, and was not mentally retarded. However she had hyperactive behavioural problem in school and at home.

There was no family history of any vascular or pigmentary disorder.

Differential diagnoses

The child has extensive naevus flammeus mainly on the right side involving the right ophthalmic branch of the fifth cranial nerve, the right upper and lower limbs and the right trunk (Figure 1). The right-sided upper lip and upper hard palate were also affected. She also has extensive aberrant Mongolian spots over a large part of the trunk and all four limbs. There were bilateral oculocutaneous melanosis (naevi of Ota) (Figure 2). A mild degree of soft tissue hypertrophy of the affected limbs was also noted.

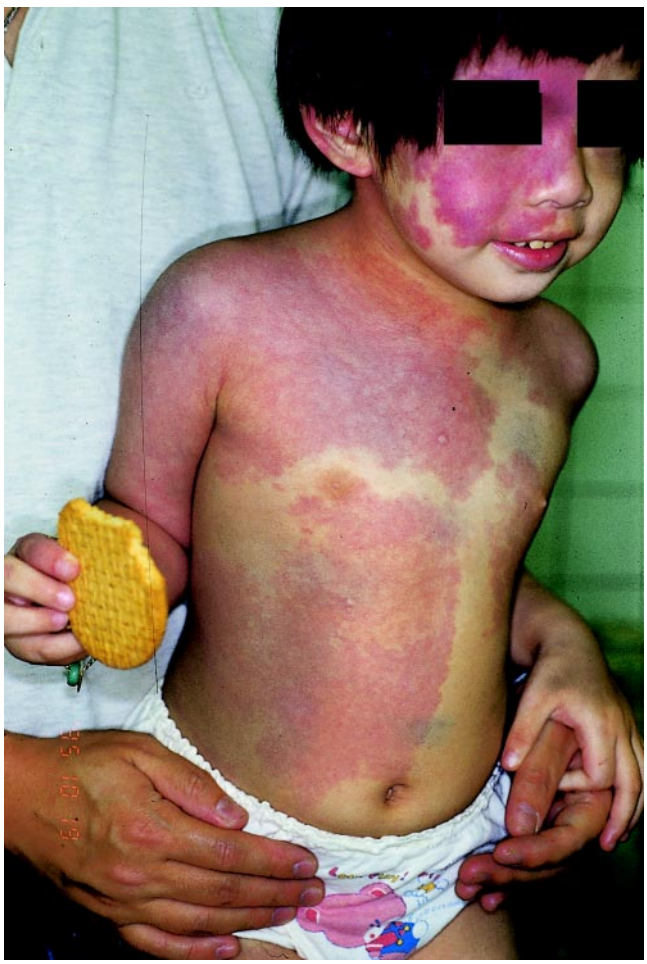

Figure 1: Naevus flammeus involving right side of face and trunk and right upper limb

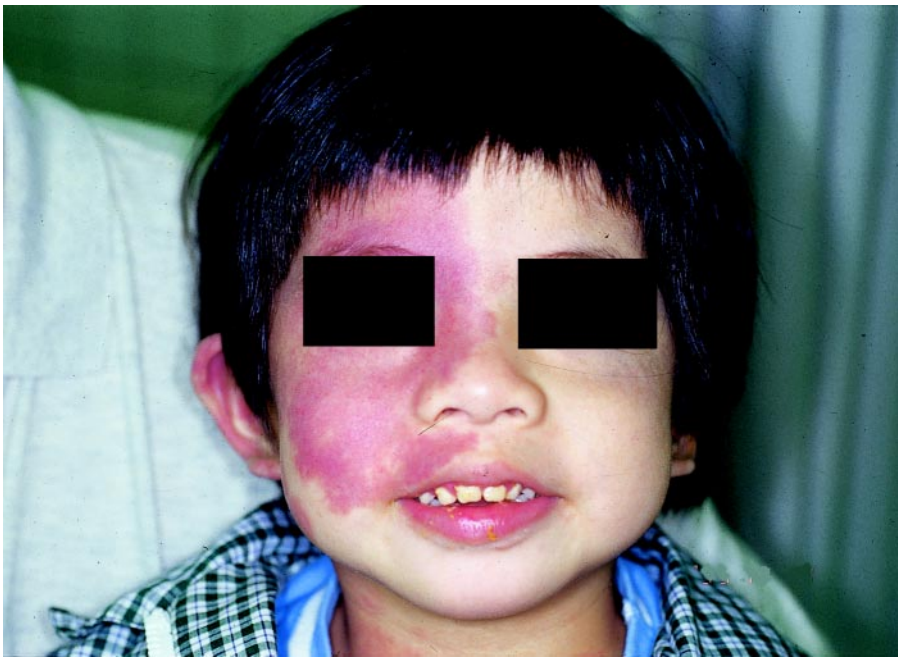

Figure 2: Naevus flammeus involving the right ophthalmic and maxillary branches and bilateral navei of Ota

Diagnosis

In summary, this child has Sturge-Weber syndrome, aberrant Mongolian blue spots and bilateral naevi of Ota, and fulfills the diagnosis of **Phakomatosis Pigmentovascularis Type IIb.**

Discussion

Question was raised whether laser therapy should be given to the patient. In general treatment result of Port Wine Stain with laser is better if performed under two years of age. Children of this age group have a poorer recollection of painful events. They will not be phobic to the procedure in the future. However, in view of the wide extent of the PWS, presence of an incurable cerebral haemangioma and the need of general anesthesia, laser therapy was considered as not suitable for her at the moment. She would nevertheless be referred to the child psychiatrist for her behavioural problem.

REVIEW ON PHAKOMATOSIS PIGMENTOVASCULARIS (PPV)

The condition was first described by Ota et al in 1947. It is a syndrome characterized by the simultaneous occurrence of a pigmented naevus and a vascular naevus in the same individual. So far, there are at least eighty cases being reported up to now, appearing mostly in the Japanese literature. Most cases have been sporadic with no significant family history. Four clinical types have been described.[1]

Classification of Phakomatosis Pigmentovascularis

Type I	naevus flammeus	naevus pigmentosis et verrucosus
Type II	naevus flammeus	aberrant Mongolian spots.
Type III	naevus flammeus	naevus spilus
Type IV	neevus flammeus	naevus spilus and aberrant Mongolian spots

They are further subdivided into (a) localized or (b) systemic, on the basis of the presence or absence of systemic involvement. Naevus anaemicus can be an additional feature in type II, III and IV. So far, there is no reported cases of systemic involvement in type I. Type II is the most common variant of PPV (up to 85% in one series).

Pathogenesis

The pathogenesis of this condition is still unclear. It may be the result of developmental abnormalities of the neural crest where vasomotor nerves and melanocytes are derived from.[2] Abnormalities in the neural regulation of blood vessels may be important in the development of vascular component of PPV. Anomalies in the migration of neural crest-derived melanocytes may result in naevus of Ota, naevus spilus, or Mongolian spot.

Differential Diagnoses

Sturge-Weber syndrome, Mongolian spots and naevus of Ota enter into the differential diagnoses of PPV.

In Sturge-Weber syndrome, the vascular malformation is usually unilateral, ended sharply at the midline, and involving part of the distribution of the trigeminal nerve. Ultrastructurally, the endothelial cells in capillaries of patients with PPV are bulkier than those in patients with Sturge-Weber syndrome, in which capillary ectasia can be observed.[3]

The naevi of Ota and Ito and Mongolian spots are lesions that histologically show dermal melanocytosis similar to the pigmentation in PPV. The epidermal basal layer is not hyperpigmented, and dermal melanocytes are present in the middle and deep dermis. In naevus of Ota unassociated with PPV, the epidermal melanocytes are increased in number, and dermal melanocytes are present in the papillary and superficial dermis.

Clinically the bluish grey spots of PPV are located all over the skin and tend to persist for life. In the autosomal dominant type of Mongolian spots as observed in healthy child, it rarely involves more than 50% of the skin and usually disappears between the ages of 4 to 6 years.

Anomalies reported in association with PPV

A number of anomalies have been reported in association with PPV. The best documented one is glaucoma. Teekhassenee-C et al [4] studied nine patients with combined oculodermal vascular malformations and oculodermal melanocytosis. They found that when oculodermal melanocytosis and naevus flammeus occur together extensively, there is a strong predisposition for congenital glaucoma. When one or both are present with partial involvement, elevated intraocular pressure may develop in later life.

Two patients with PPV type IIb were reported to have iris mammillations.[5] The association of melanosis oculi and iris mamillations may reflect a disorder of migration or development of neural crest-derived elements.

One patient with PPV was also reported to have selective IgA deficiency,[6] while another patient was reported to have multiple granular cell tumour.[7] Whether these occur by chance or are genuine associations remain unknown.

Learning points:

Phakomatosis Pigmentovascularis describes developmental anomaly characterized by simultaneous occurrence of vascular and pigmented naevus, with associated eye abnormalities.

References

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