Phacomatosis Cesioflammea Associated with Pectus Excavatum

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Sir,

Phacomatosis cesioflammea is a rare genetic disease characterized by the coexistence of a blue spot (such as an aberrant Mongolian spot), a naevus flammeus, and sometimes a naevus anaemicus. This disorder is often associated with neurological and skeletal abnormalities. We report here a 9-month-old infant boy with this rare cutaneous disease presenting a depressed-chest deformity. To the best of our knowledge, this is the first report of pectus excavatum as a possible musculoskeletal manifestation in phacomatosis cesioflammea.

CASE REPORT

A 9-month-old male infant, who had been born at full-term, presented for an evaluation of congenital skin lesions. He was the first child born to unrelated, normal-appearing parents. His family history was unremarkable, and he had no known relatives with phacomatosis cesioflammea.

The child presented with reticulated vascular lesions on his trunk and limbs (Fig. 1A). These marble-like telangiectasia became more apparent when he cried or after physical exertion. The cyanosis in his left leg became more profound when he was irritated. In addition, there was a large poorly defined patch of bluish-grey pigmentation on his back and sacral area (Fig. 1B). Physical examination revealed congenital pectus excavatum (Fig. 1A) and no evidence of respiratory distress. No remarkable neurological deficits or growth or developmental delays were observed at 9 months of age.

DISCUSSION

Phacomatosis cesioflammea is a variant of phacomatosis pigmentovascularis (PPV), a genetic disease characterized by the presence of two or more naevi. Hasegawa classifies PPV into four types (1). Each type involves a naevus flammeus with an additional naevus and is further divided into subtypes a and b. Subtype a has cutaneous involvement, and subtype b extracutaneous involvement.

Cutaneous marmorata telangiectasia congenital (CMTC) with Mongolian spots has recently been reported as type V, a distinct type of PPV (2). In 2005, Happle (3) proposed replacing PPV types II, III and V with the terms phacomatosis cesioflammea, phacomatosis spilorosea, and phacomatosis cesiomarmorata (Table I). The concept of capillary malformations includes some different clinical presentations, and not all authors agree on a denomination, especially in the case of reticulated capillary malformations, which can be very difficult to differentiate from CMTC.

In the present case, the infant had diffuse reticulate capillary malformation associated with large Mongolian spots. Capillary malformation, which usually presents as a port-wine stain or naevus flammeus, is the most common type of vascular malformation. As a congenital malformation of the superficial dermal blood vessels, capillary malformation is present at birth, grows in size along with the child's growth, and remains for life.

A variety of extracutaneous abnormalities have been associated with capillary malformation, and several syndromes have been delineated. Sturge-Weber syndrome is characterized by the triad of capillary malformations involving the upper facial dermis, the ipsilateral leptomeninges, and the ipsilateral cerebral cortex. Klippel-Trenaunay syndrome manifests as a triad of capillary

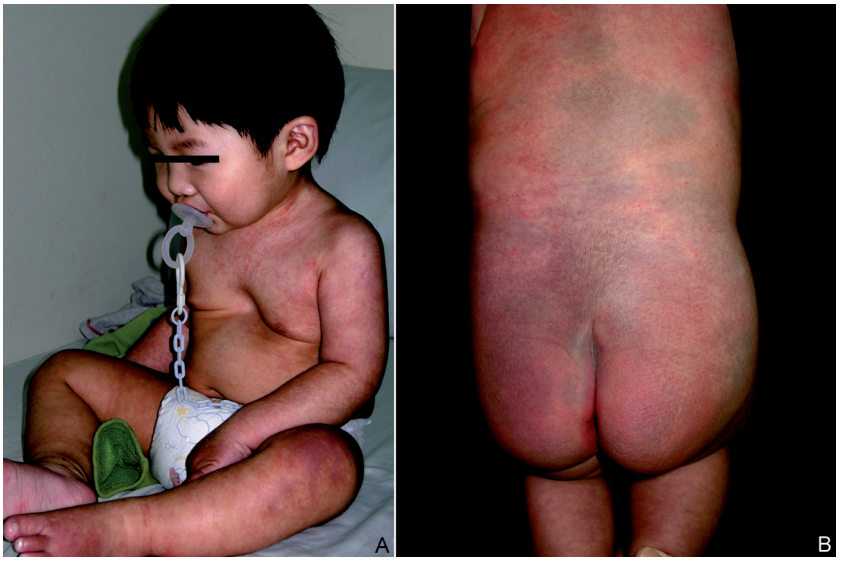

Fig. 1. (A) Extensive areas of reticulated, marble-like erythema over the patient's limbs and chest. Also note pectus excavatum. (B) Large area of poorly defined bluish-grey pigmentation distributed over the patient's upper back and extending to the sacral area, partially mixed with diffuse reticulate capillary malformation.

Table I. *Comparison of the traditional and new classifications of phacomatosis pigmentovascularis (PPV)*

Traditional classification (1)	Vascular component	Pigmentary component	New classification (3)
I a/b	Naevus flammeus	Pigmented linear epidermal naevus	No PPV
II a/b	Naevus flammeus ± naevus anaemicus	Blue spots	Cesioflammea
III a/b	Naevus flammeus ± naevus anaemicus	Naevus spilus	Spilorosea
IV a/b	Naevus flammeus ± naevus anaemicus	Blue spots and naevus spilus	Unclassifiable
V a/b	Cutis marmorata telangiectasia congenital	Blue spots	Cesiomarmorata

malformation, congenital varicose veins, and hypertrophy of underlying tissues, particularly skeletal overgrowth. Similarly, Parkes-Weber syndrome comprises an arteriovenous (AV) malformation in addition to the triad of Klippel-Trenaunay syndrome. Cobb syndrome is characterized by a cutaneous vascular lesion in the skin overlying the spine in association with vascular malformation in the subjacent spinal meninges. Wyburn-Mason syndrome (unilateral retinocephalic syndrome) manifests as facial capillary malformations associated with unilateral AV malformation of the retina and the intracranial optic pathway. However, our case did not fit any of these known capillary malformation syndromes.

The simultaneous occurrence of capillary malformations and Mongolian spots led to the diagnosis of PPV in our patient. In PPV, the presence of capillary malformations may easily be confused with CMTC. However, a true CMTC involves depressed furrows and linear cutaneous atrophy over the joints, which are not seen in capillary malformation. Therefore this patient was diagnosed with PPV type II, or phacomatosis cesioflammea.

Capillary malformation overlying the lumbar spine may be a marker for an underlying primary skeletal or neurological abnormality such as spinal dysraphism, tethered spinal cord, lipomenigocele, or diastematomyelia. However, the skeletal deformity in the present case was at the front, instead of corresponding to the capillary malformation lesion at the back. It has been proposed that phacomatosis cesioflammea is an example of didymosis, indicating the coexistence of different naevi in the same patient (4). Some authors have hypothesized that an embryogenesis error in early gestation involving the migration of mesoderm and ectoderm may affect foetal vascular development and melanocytic migration to cause this condition. In our patient, who presented with pectus excavatum, a funnel-shaped deformity in the anterior chest wall involving the sternum and costal cartilage might have occurred through this mechanism. Although the exact aetiology is unknown, the overgrowth of costal cartilages that rotate and curve dorsally at the early embryonic stage may be the cause of this syndrome (5).

Table II. *Skeletal abnormalities associated with phacomatosis pigmentovascularis (PPV)*

Type of PPV/ New classification	Ref.	Musculoskeletal abnormalities
Type IIb/ Cesioflammea	(7)	Hemifacial and unilateral leg hypertrophy, tibial bone cyst, scoliosis, syndactily of 3rd and 4th digits
	Our case	Pectus excavatum
Type IIIb/ Spilorosea	(8)	Convex scoliosis
	(9)	Difference in size of the extremities
	(10)	Short stature
Type Vb/ Cesiomarmorata	(2)	Hypertrophy of left buttock and thigh

The extracutaneous manifestations of PPV range from neurological abnormalities and ocular alterations to involvement of a variety of musculoskeletal and internal organs, including skeletal abnormalities. Anomalies such as discrepancy in the length of extremities, scoliosis, spinal dysraphism, hemihypertrophy, syndactilia, and premature eruption of the teeth (6) have been described (Table II). In the present case, the pectus excavatum and skeletal anomalies may be either a random occurrence or a possible manifestation of the disease. Additional case reports of phacomatosis cesioflammea would help further our understanding of this disorder.

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