A Case of Phacomatosis Pigmentokeratotica in Japanese Monozygotic Twins

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Epidermal naevus syndromes include different diseases that share a common feature of mosaicism (1, 2). The association of a speckled lentiginous naevus, an organoid naevus with sebaceous differentiation, with skeletal and neurological anomalies, constitutes a specific syndrome called phacomatosis pigmentokeratotica (PPK).

It has been hypothesized that the co-occurrence of two different naevi reflects a so-called twin spot phenomenon (3). We report here an unusual example of PPK in a pair of Japanese monozygotic twins, associated with multiple pinhead-sized vascular lesions, aortic stenosis, arteriovenous fistulation and hypertrophy of the ipsilateral leg.

CASE REPORT

A 25-year-old woman, who was a monozygotic younger twin (with a healthy female twin), had had yellow papules linearly arranged along Blaschko's lines on the left half of the body, excluding the face and head, at birth (Fig. 1). At 25 years of age, a mass was detected in her left breast, and she was referred to our hospital for examination and treatment. She had pale brown spots on the left back in the precordial region, and erythematous macules with crusting were present above these spots. Multiple papules that were normal in colour with a warty surface were present within these macules. Aggregations of similar papules were also observed in the posterior neck-left shoulder, left back, left upper limb, and the lumbar area, showing characteristic distribution along Blaschko's lines. Multiple pinhead-sized vascular lesions were found within these hyperpigmented areas (Fig. 2).

A skin biopsy of a papule from the left back demonstrated matured sebaceous glands forming nests in the dermis, some of which were open to the epidermis, along with immature hair follicles. Based on these findings, a diagnosis of sebaceous naevus was made.

On contrast-enhanced computed tomography (CT) of the chest, arteriovenous fistulation was detected in the left thoracic wall, centred on the left 4th intercostal space, and dilation of the left thoracic arteries/veins was also noted (Fig. 3). The mass lesion in the left breast detected on medical examination was thought to be the result of this arteriovenous fistulation. Magnetic resonance angiography demonstrated slight stenosis of the descending aorta, while X-ray examination of the legs showed left coxarthrosis and impaired longitudinal bone growth

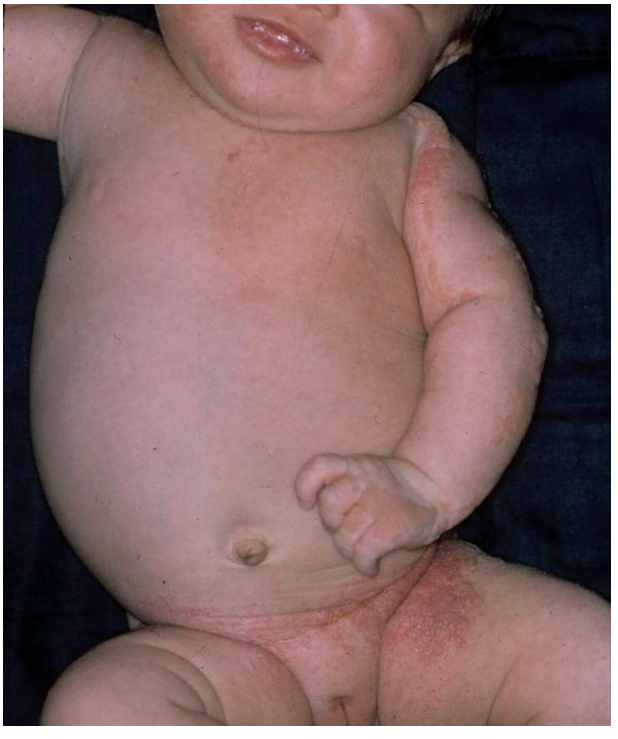

Fig. 1. Clinical appearance (40 days after birth). Reddish-yellow warty eruptions accompanied by discharge were observed along Blaschko's lines on the left back in the brachium, left precordial region, and inguinal region.

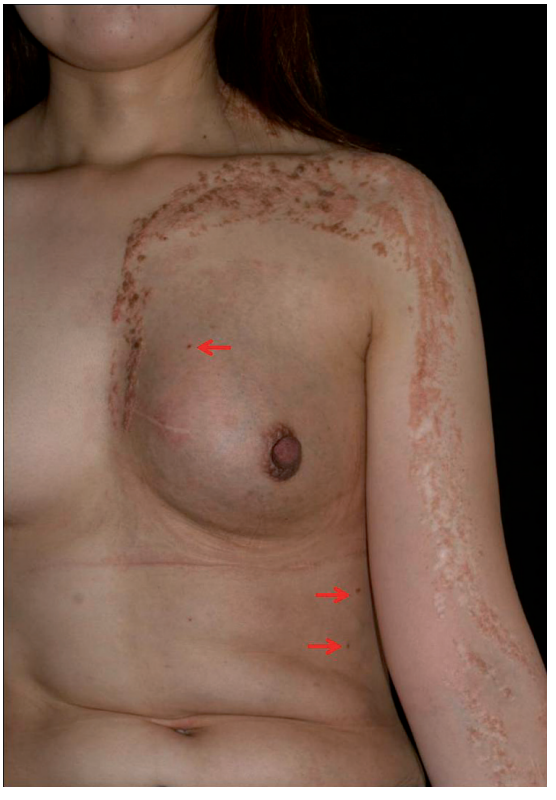

Fig. 2. Clinical appearance (at 25 years of age). Along Blaschko's lines of the left trunk and all limbs, there were multiple warty eruptions that were normal in colour to slightly yellowish with crusts. There were scattered, dilated veins and multiple pinhead-sized vascular lesions (arrows) in the left precordial region.

in the left leg. The skeletal and vascular anomalies were limited to the left half of the body with skin lesions. Neurological and ophthalmological explorations gave normal results.

DISCUSSION

PPK is delineated by the presence of multiple organoid naevi (ON) with sebaceous differentiation, arranged according to Blaschko's lines, along with a speckled lentiginous naevus (SLN) of the papular type arranged in a chequerboard pattern. The two types of naevi are predominantly ipsilateral or contralateral, but may also be bilaterally distributed (4). Other anomalies, usually neurological, skeletal or ophthalmological, include hemiatrophy with muscular weakness, segmental dysaesthesia and hyperhidrosis, mild mental disability, seizures and deafness, ptosis and strabismus (5).

The most important differential diagnosis is Schimmelpenning syndrome, a disorder that is likewise characterized by ON with sebaceous differentiation in association with ocular, neurological and skeletal defects. In PPK, neurological defects, including mental deficiency, are often present, but are less severe than those noted in Schimmelpenning syndrome, whereas the characteristic neurological features of the associated papular naevus spilus syndrome, such as hyperhidrosis, muscular weakness, dysaesthesia, or sensory or motor neuropathy, may also be found (1).

Moreover, in PPK, other typical findings of Schimmelpenning syndrome, such as coloboma and lipodermoid of the conjunctiva, have been absent from cases reported to date.

Although our patient did not demonstrate any small dark speckles in the form of melanocytic naevi within the café-au-lait hyperpigmentation, the large unilateral macule on the left side of the trunk with a strict midline separation and lack of neurological defects, such as mental deficiency and ophthalmological defects, are rather considered part of PPK. Multiple pinhead-sized angioma-like lesions superimposed on SLN in PPK have been described by Tadini et al. (5) and Boente et al. (6). In our patient, such pinhead-sized vascular lesions were present within the hyperpigmented area. Such vascular lesions are rather typical of PPK.

Other vascular abnormalities, such as aortic coarctation, renal artery stenosis, dilation or stenosis of the pulmonary artery, valvular stenosis or insufficiency, are sometimes seen in Schimmelpenning syndrome (7). Boente et al. (6) reported a case of PPK with aortic stenosis, and considered that such vascular defects might be a component of Schimmelpenning syndrome representing one-half of the PPK process. These vascular anomalies were strikingly similar to those of our patient, but the arteriovenous fistulation noted in our patient, to our knowledge, has not been reported previously in patients with PPK.

All cases reported to date have been sporadic. PPK is an example of an autosomal recessive mutation that is usually lethal but is rescued by mosaicism (8). In our patient, there were no findings of skin symptoms suggesting other types of naevus in her twin sister, which supports the theory of mosaic mutation.

To date, approximately 30 cases of PPK have been reported. We propose that the present case represents an unusual example of PPK.

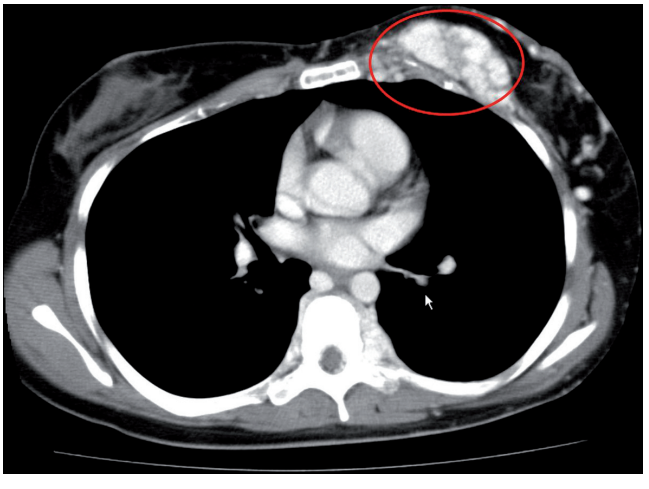

Fig. 3. Contrast-enhanced computed tomography (CT) of the chest. Arteriovenous fistulation was detected on the left thoracic wall centring on the left 4th intercostal space.

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