

Familial multiple naevi flammei

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SUMMARY The third family with multiple naevi flammei and the first description of its occurrence in identical twins is presented. Autosomal dominant inheritance is confirmed.

Naevus flammeus, also known as port wine stain, is a birthmark found in 0.3 to 0.5% of newborn infants.¹ These naevi are not generally considered hereditary. However, two descriptions of a family with genetically determined multiple naevi flammei have been reported.^{2,3} We present another family indicating autosomal dominant inheritance with variable expression of multiple port wine stains.

Description of family

The proband, a 4 year old girl, the first child of healthy unrelated Ashkenazi Jewish parents, was referred to our clinic because of upper respiratory infections. Her past medical history was unremarkable. Physical examination revealed no abnormalities except for five naevi flammei, two on her neck, two on her upper extremities, measuring from 0.5 to 4.0 cm in diameter, and a very large one, purple in colour, on her right groin and upper part of her leg. She had two sibs, non-identical twins, one with a large naevus on the temporal region and several on the upper extremities and the other with naevi on the forehead, back, and extremities. The mother remarked that similar birthmarks were common in their family. She herself had six naevi flammei, with diameters of 0.5 to 1.5 cm, on her upper and lower limbs and another member of the family, an aunt, had naevi on the extremities and chest. The family pedigree is shown in the figure.

In all the family members examined by us the naevi flammei were bilateral, varying in size and location as well as in colour, which ranged from pink to purple. The lesions on the head tended to be purple while those on the extremities were generally pink to red. In the adult patients the lesions were smaller than in the children. None of the family members had any evidence of other haemangiomas, malformations, or visceral involvement. The

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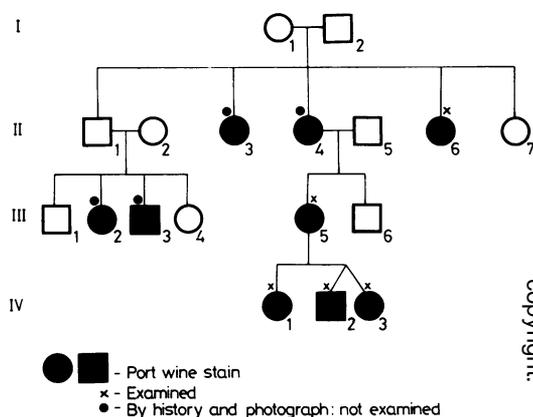


FIGURE Pedigree of the family with multiple naevi flammei.

children's father and his family were healthy with no naevi.

Discussion

The port wine stain is a vascular naevus composed of dilated mature capillaries in the dermis.¹ The stain is sharply circumscribed, ranging from a small red macule to a large patch and varying in colour from pink to bluish red.⁴ It is present at birth and shows no tendency to resolve, in contrast to the salmon patch, a lesion which is sometimes erroneously called naevus flammeus.^{1,4} The cause of port wine stains is unknown; it is a developmental defect which is not considered to be hereditary.⁵ Most naevi flammei occur as isolated defects. However, they may occur in certain vascular syndromes, such as the Sturge-Weber syndrome.

In 1949 Shelley and Livingood² described the pedigree of a family with multiple naevi flammei.²

In 1968 a family with naevi flammei of the forehead was described.³ Apart from these pedigrees, we could find no other published descriptions. The characteristics of the pedigree under study point strongly to autosomal dominant inheritance: there is a vertical pattern of transmission, one of the parents is affected, the expressivity is markedly variable, and there is no consanguinity. A previously undescribed feature is the occurrence of naevi in non-identical twins. It appears that naevi flammei can be isolated, syndromic, or follow autosomal dominant inheritance. Although the last pattern has rarely been reported it is possible that with awareness of its existence more families will be found.

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