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Case Report

Hereditary benign telangiectasia – first family in Northern Ireland

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Hereditary benign telangiectasia (HBT), first described in 1971,[1] is a rare autosomal dominant disorder.[2, 3, 4] It is characterised by widespread telangiectases which may be punctate, plaque-like, radiating or merely a diffuse blush. The lesions generally are observed before adolescence, and rarely during the first year of life.[5] It is important to recognise HBT as it may only cause cosmetic disability and usually is not associated with any significant clinical disturbance. This contrasts with hereditary haemorrhagic telangiectasia (Rendu-Weber-Osler disease; HHT), in which there is a major risk of severe haemorrhage in adults.[6, 7] We describe the first family with HBT in Northern Ireland and discuss the differentiation from other primary telangiectatic disorders.

CASE REPORT A 10-year-old girl was referred by her family doctor because of telangiectases affecting the arms, face and legs. The lesions first appeared at the age of 5 years. During the past year the lesions had become more numerous. There was no history of epistaxis, jaundice, liver disease or connective tissue disorders. On examination, small telangiectases were noted over hands (Fig.), arms, upper trunk and face. In total, there were 13 telangiectases, 0.5 mm in size on average. Each lesion was flat and non-tender. The mucous membranes were not involved. There were no other abnormal clinical features.

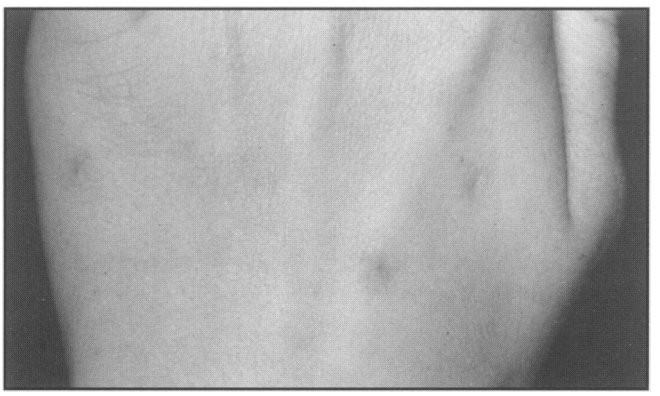

Figure 10-year-old girl: hand showing several telangiectases.

The patient was the eldest of a sibship of three; she has a brother (aged 8 years) and sister (aged 6 years). The brother had three telangiectases on his face and arms. The sister also had a few telangiectases. The mother had no skin abnormality but the father had telangiectases of his hand since childhood. The paternal grandmother had several lesions which also had first appeared in childhood.

DISCUSSION

Telangiectasia is due to the persistent dilatation of pre-existing small blood vessels, usually in the skin or mucous membranes. Primary telangiectasia includes HBT, HHT, generalised essential telangiectasia and spider angiomas. Generalised essential telangiectasia is an

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uncommon disorder which usually has its onset in middle age.[8] Widespread sheets of linear telangiectasias occur on the legs, arms and abdomen.[4] It is more frequently seen in women and is not associated with spontaneous bleeding.[8] A spider angioma morphologically is a red spot from which small legs radiate out. Such lesions are often surrounded by halo of pallor.[9] They occur in healthy individuals but are associated with pregnancy, oral contraceptive pill use and hepatic dysfunction.[4] Secondary telangiectasia occurs as a consequence of a known disease such as connective tissue diseases and in cutaneous conditions such as acne rosacea.[4] Few cases of hereditary benign telangiectasia have been reported.[1-5] It may be because of the benign nature of HBT, as it often goes unrecognised. However, it is important to distinguish HBT from the other primary and secondary telangiectasia, especially HHT, which is a chronic debilitating disease of vascular malformation also transmitted as an autosomal dominant trait.[6, 7] Three types of vascular malformation are found in HHT, namely telangiectases, arterio-venous malformations and aneurysms. The characteristic telangiectasia of HHT is a 1-3 mm red to violet punctate lesion which is sharply demarcated from the surrounding skin, usually found on the face, lips, nares, tongue, ears, hands, chest and feet. The telangiectases often increase in size and number with age.[6,7] The arteriovenous malformations and aneurysmal dilatations of large arteries, which may occur in HHT are not seen in HBT and this can be used to distinguish HBT from HHT. However, as Ryan and Wells state in their original description "there is nothing unique about the pattern of telangiectasias in hereditary benign telangiectasia".

The pathogenesis of HBT is unknown. It is due to an autosomal dominant gene.[1-5] The only treatment that may be required is electrocautery or laser therapy for cosmetic reasons. It is important to be aware of HBT as a cause of generalised telangiectasia and that it is a benign disorder.

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