

## Hereditary benign telangiectasia: image analysis of hitherto unknown association with arteriovenous malformation

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### Summary

We report 10 patients with hereditary benign telangiectasia (HBT), in whom the age ranged from 1 to 19 years (mean 7.5). The male/female ratio was 1 : 2.3 (3 : 7). Four patients (40%) had congenital lesions. Image analysis with Doppler echogram, angiography and thermography revealed an arterial component in the lesions, consistent with arteriovenous malformation (AVM). This is the first report of HBT in association with AVM.

*Key words:* angiography, arteriovenous malformation, Doppler echogram, haemangioma, hereditary benign telangiectasia, thermography

Ryan and Wells<sup>1</sup> first described hereditary benign telangiectasia (HBT) as a new clinical entity in 1971. They reported seven kindred with HBT and defined characteristic features of the disease as follows: (i) telangiectasia appears after the first year of life and often before adolescence; (ii) the lesions are punctate, plaque like, radiating, arborizing, reticulated, mottled, spider-like or merely a diffuse blush; (iii) the lesions are more prominent during pregnancy; (iv) there is an absence of associated disease; (v) the main pathological feature is dilatation of the horizontal subpapillary venous plexus; and (vi) inheritance of the trait is likely to be autosomally dominant. Although several cases of HBT have been reported,<sup>2–10</sup> the aetiology of the disease is still obscure.

We have noted local increased temperature in association with faint but notable arterial pulsation of several parts of the HBT lesions in our 10 patients. These findings have never been reported previously. We have therefore evaluated the HBT lesions with several imaging procedures such as Doppler echogram,

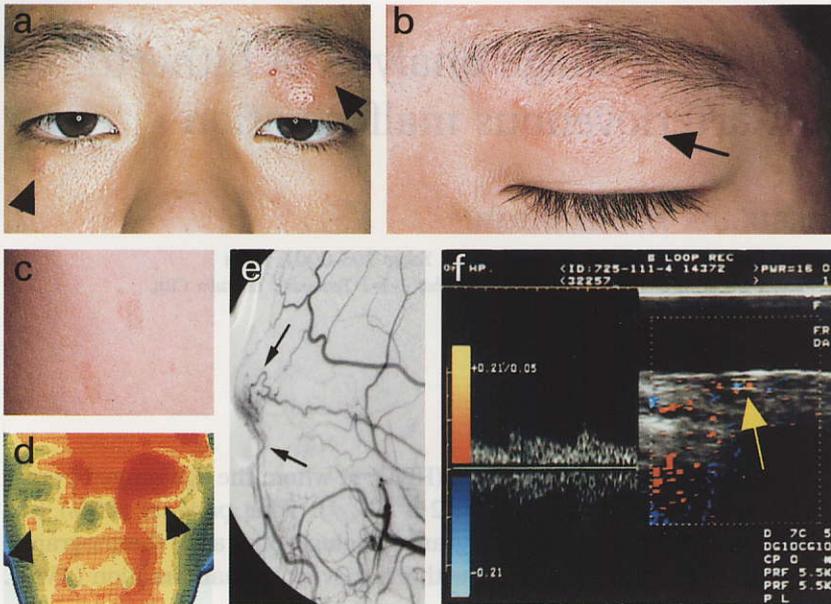
angiography and thermography, in order to identify the arterial component in the HBT lesions.

### Case reports

#### Patient 1

A 16-year-old boy had congenital multiple reddish lesions on his upper left eyelid, lower right eyelid and upper left arm. He had recently noticed slight growth of these lesions (Fig. 1a–c) and was referred to us for treatment. His past history was unremarkable. His younger brother also had multiple reddish plaques on the brow and the upper arm (Fig. 2a–c), and his father had a similar lesion on his lower back (Fig. 2d). Physical examination of the proband revealed a reddish-brown and slightly elevated lesion on the upper left eyelid, and multiple reddish macules and patches on the lower right eyelid and upper left arm. Some on the upper left eyelid and lower right eyelid were elevated and dark coloured, and the rest were flat and light coloured with a mottled configuration. On palpation, arterial pulsation and local heat were noted in the lesion on the upper left eyelid. Thermography of the face revealed a localized high temperature (Fig. 1d, arrowed). Digital subtraction angiography and Doppler

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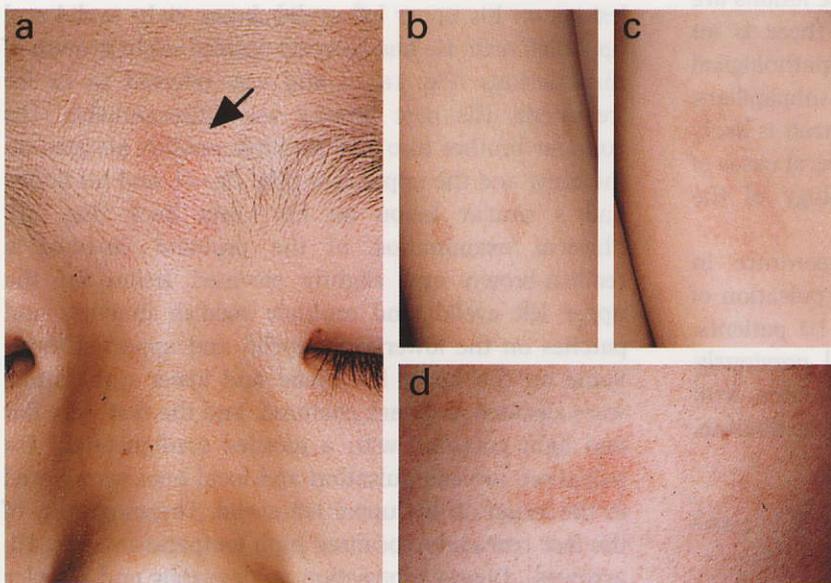
**Figure 1.** Patient 1. (a–c) Clinical appearance: a reddish-brown and slightly elevated pulsatile lesion is evident on the upper left eyelid (arrow), and multiple reddish macules and patches on the lower right eyelid (arrowhead) and upper left arm. (d) Thermography of the face revealed localized high temperature (arrowheads). (e,f) Digital subtraction angiography (e) taken against the left eyelid detected the feeding arteries (arrows). Doppler echogram revealed remarkable arterial blood flow (f, left: arterial pattern) in the lower dermis detected from the area between the yellow double line (f, right, arrow), which place corresponds to the medial margin of the lesion. Such findings were not observed on the unaffected right eyelid.

echogram against his left eyelid detected feeding arteries (Fig. 1e,f, arrowed), early venous filling and late phase pooling of contrast medium, consistent with the findings of arteriovenous malformation (AVM). Histology of a biopsy from the left eyelid showed thick-walled arteries in the lower dermis (Fig. 3a,c), ascending into the upper dermis in a spiral fashion. In the upper dermis (Fig. 3b), proliferation of irregularly shaped aberrant blood vessels was prominent, with a lymphocytic inflammatory infiltrate. The diagnosis of HBT associated with AVM was made. Combination therapy with

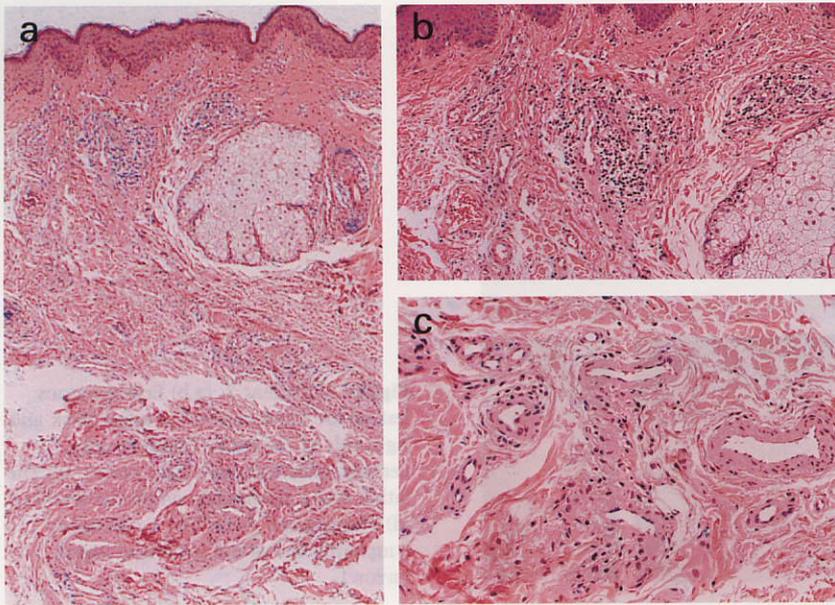
selective arterial embolization, pulsed dye laser and scalpel surgery was performed.

*Patient 2*

A 2-year-old girl had a reddish mark on her upper right arm. Her mother also had small erythematous patches on her right forearm and abdomen. Physical examination of the proband revealed an irregularly shaped erythematous lesion, speckled in colour and the margin of which faded into a white halo (Fig. 4a). A small telangiectatic lesion was also found on the left auricle



**Figure 2.** Clinical appearance of relatives of patient 1. (a–c) The younger brother had multiple reddish plaques on the brow (arrow) and the upper arms. (d) The father had an erythematous patch on the lower back.



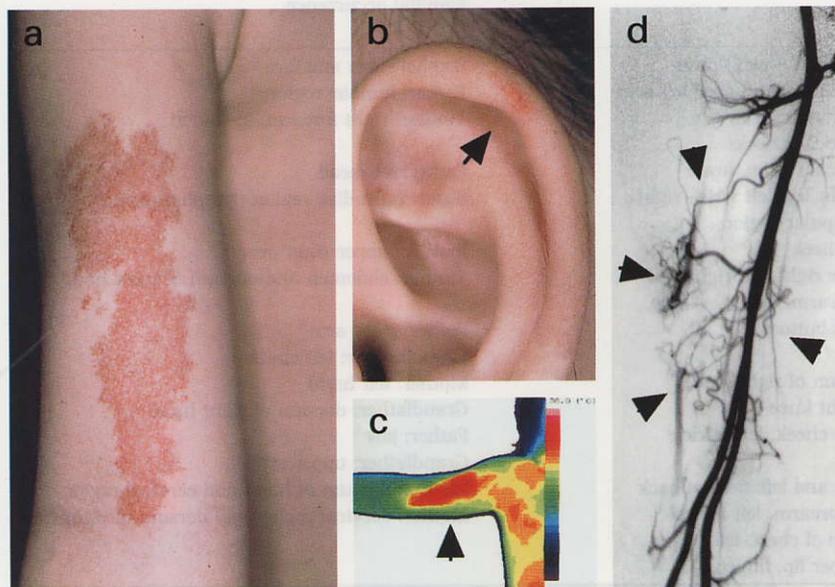
**Figure 3.** Patient 1: histological findings. (a,c) Thick-walled arteries in the lower dermis ascend into the upper dermis in a spiral fashion. (b) In the upper dermis, note proliferation of irregularly shaped aberrant blood vessels. The inflammatory infiltrate is composed mainly of lymphocytes (haematoxylin and eosin; original magnification: a,  $\times 40$ ; b,  $\times 100$ ; c,  $\times 200$ ).

(Fig. 4b). Thermography and angiography demonstrated localized heat and typical findings of AVM (Fig. 4c,d), as in patient 1. We totally resected the lesion with ligation of the feeding arteries. Four years after surgery, multiple new lesions with a spider-like appearance developed on her cheeks.

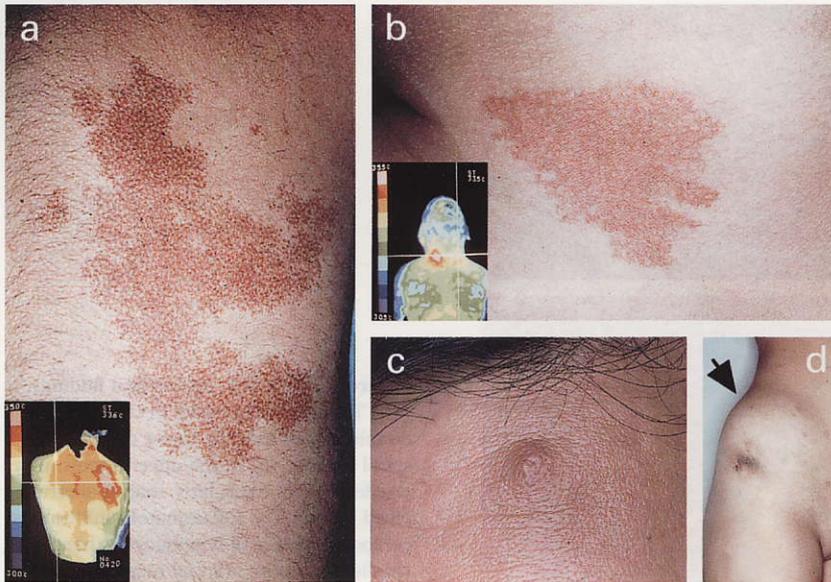
#### Patient 3

A 4-year-old girl had developed reddish plaques on many parts of her body since she was 1 year old

(Fig. 5a,b). Her first visit was in 1982. Her father had had a small compact haemangioma on the forehead since infancy (Fig. 5c), and her younger sister developed a cavernous lymphangioma at the right iliac region (Fig. 5d, arrowed). Physical examination of the proband revealed multiple erythematous lesions on her upper back, right side of the neck, left forearm and lower right leg, all of which showed irregularly shaped reddish-brown plaques with local heat. Thermography vividly demonstrated the high temperature of the lesions (Fig. 5a,b).



**Figure 4.** Patient 2. (a,b) Clinical appearance: an irregularly shaped erythematous lesion is evident on the upper right arm and a small telangiectatic lesion is seen on the left auricle (arrow). (c) Thermography detected localized heat (arrow). (d) Angiography demonstrated feeding arteries (arrowheads).



**Figure 5.** Patient 3. (a,b) Erythematous lesions are apparent on the upper back and right side of the neck. Thermography demonstrated increased heat (inset). (c) The father had a haemangioma on the forehead. (d) The younger sister had a cavernous lymphangioma at the right iliac region (arrow).

## Discussion

HBT is a rare familial disorder manifesting as multiple cutaneous haemangiomas or telangiectatic lesions. Although several cases of HBT have been reported,<sup>2-10</sup> the aetiology of the disease is still obscure: angiogenetic factors or hypersensitivity of oestrogen and progesterone receptors in the affected lesions have been suggested.<sup>6</sup>

We have seen 10 patients with HBT over 18 years (Table 1), of whom the age ranged from 1 to 19 years (mean 7.5). The male/female ratio was 1 : 2.3 (3 : 7).

The family history was positive in all cases. In four patients (40%), lesions had been noticed at birth. No patient had associated disease. The clinical features were very characteristic. The colour of the lesions was not uniform but was very varied, showing reticular, mottled, spider-like, speckled or plaque-like features with a faded margin or white halo. Although the clinical appearance was sometimes similar to that of a port-wine stain (PWS), all of our cases of HBT showed prominent local heat, unlike a PWS.

We also found faint but notable arterial pulsation on

**Table 1.** Patients with hereditary benign telangiectasia

Patient no.	Sex/age (years)	Site	Familial occurrence
1	M/16	Upper left eyelid, lower right eyelid, upper left arm	Brother: arm and face Father: lumbar region
2	F/2.5	Upper right arm, left auricle, cheeks	Mother: right forearm, abdomen
3	F/4	Cervical region, lower right leg, left wrist, right scapular region	Father: forehead Sister: right iliac region (lymphangioma)
4	F/1	Left cheek	Mother: upper right arm
5	F/1.7	Lower right leg, right forearm, upper left arm	Mother: abdomen and dorsum of right hand
6	F/2	Right buttock	Father: right arm Grandmother: left cheek
7	M/6	Dorsum of right foot, right knee	Mother: left hand Grandfather: dorsum of right hand
8	M/9	Right cheek, left auricle	Father: jaw Grandfather: upper arm
9	F/19	Right and left sides of back	Mother: dorsum of hand and cervical region
10	F/14	Left forearm, left lateral side of chest, left palm, upper lip, finger	Mother: cervical region and dorsum of right hand

Careful palpation. The pulsation was usually localized to one or several parts of the margin of these lesions. These findings have not been reported previously. As they were suggestive of an associated AVM, we analysed the lesions by imaging devices. Our patients differed from those with ordinary AVM in their family history, the multiplicity, flat appearance and localization of arterial pulsation in their lesions, and stability of their clinical course. Thermography does not show such an elevated temperature in lesions of venous haemangioma, including PWS. As presented above, this imaging study clearly demonstrated the arterial component in HBT lesions, consistent with the findings of AVM. To our knowledge, this is the first report of HBT associated with AVM.

Unfortunately, we were not able to identify histological evidence of AVM in all our patients, but the association with an abnormal arterial supply was distinctively demonstrated on image analysis. This result also indicates difficulty with treatment. Pulsed dye laser alone had minimum effect in our patients. We believe that HBT is best treated by combination treatment with selective arterial embolization and scalpel or laser surgery.

In addition, although HBT can be clinically differentiated from ordinary PWS, it is often misdiagnosed as PWS and usually resists pulsed dye laser treatment. We consider that dye laser treatment alone is not sufficient in the treatment of haemangioma in association with AVM because of the abundant arterial blood flow. Image analysis should be done in cases of resistant 'PWS', and at the same time careful palpation and

confirmation of multiplicity and familial occurrence are strongly recommended.

The multiplicity of HBT lesions may be a result of stimulation by angiogenetic factors that are induced by AVM. We also consider that patient 3, with a family history of cavernous lymphangioma, raises the aetiological possibility of HBT as a familial lymphoangiomatous vascular malformation.

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