

# HEREDITARY BENIGN TELANGIECTASIA: A CASE PRESENTATION AND REVIEW OF LITERATURE

A. OANȚĂ<sup>1</sup> V. VEREGUȚ<sup>2</sup> D. E. BRĂNIȘTEANU<sup>3</sup>  
G. STOLERIU<sup>3</sup> M. IRIMIE<sup>1</sup>

**Abstract:** *Hereditary benign telangiectasia is a rare condition characterized by cutaneous generalized telangiectasias and angiomas. This diagnosis should be suspected in patients with this type of lesions, with a positive family history, without bleeding or mucosal involvement.*

*We present the case of a man aged 30 years with numerous telangiectasia and angiomas located on the face, neck, anterior and posterior thorax, arms, without bleeding. Histopathology revealed a normal epidermis with dilated subpapilar plexus. The anamnesis revealed similar lesions in other family members. Electrocauterization or laser therapy has been proposed as treatment of these lesions.*

*Differential diagnosis must be done mainly with other two rare conditions: hereditary hemorrhagic telangiectasia and generalized essential telangiectasia.*

**Key words:** *hereditary benign telangiectasia, hereditary hemorrhagic telangiectasia, generalized essential telangiectasia.*

## 1. Introduction

Hereditary benign telangiectasia (HBT) is a rare clinical condition described by Ryan and Wells [15]. HBT is characterized by the presence of cutaneous teleangiectasias, family history, the absence of mucosal involvement and absence of haemorrhages. We present the case of a patient diagnosed with HBT.

## 2. Clinical case

A 30-years old male patient was consulted for numerous telangiectasias and angiomas localized on the photoexposed areas such as the face, thorax and limbs. The anamnesis reveals that these lesions started to appear from the age of 10 years, initially on anterior thorax with subsequently expansion on rest of areas. Patient's father (fig. 1), grandfather and great-grandfather of paternal line, a cousin of the patient and her

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<sup>1</sup> Faculty of Medicine, *Transilvania* University of Braşov.

<sup>2</sup> Emergency County Hospital in Brasov

<sup>3</sup> University of Medicine and Pharmacy G.T. Popa of Iași.

daughter shows identical lesions.

Dermatological examination revealed telangiectasias and angiomas with dotted, round, oval or dendritic appearance,

arranged in plaques and located on the face, neck, arms, anterior and posterior thorax (fig. 2, 3, 4).



Fig. 1. *Clinical appearance of hereditary benign telangiectasia (patient's father)*



Fig. 2. *Clinical appearance of hereditary benign telangiectasia – numerous telangiectasias and angiomas on chest and arms (patient)*



Fig. 3. *Clinical appearance of hereditary benign telangiectasia – numerous telangiectasias and angiomas on posterior thorax (patient)*



Fig. 4. *Clinical appearance of hereditary benign telangiectasia –telangiectasias and angiomas on the face (patient)*

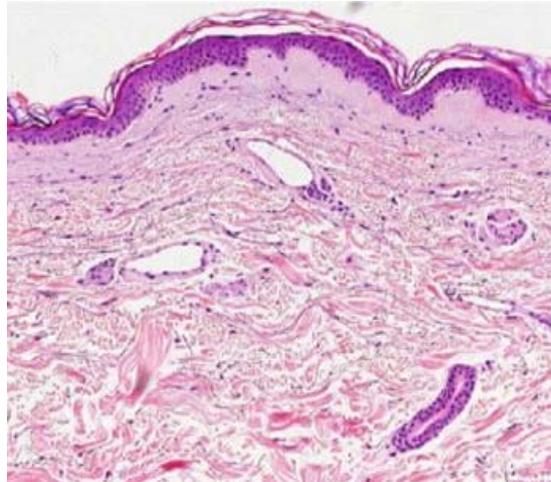


Fig. 5. *Histopathological appearance of hereditary benign telangiectasia (H&E, x40)*

The patient had not epistaxis, melaena, haematuria or cutaneous bleeding. Haematological and biochemical investigations, coagulation tests, estrogen, progesterone and testosterone levels were within normal limits. Histopathology revealed a normal epidermis with dilatation of subpapillar plexus (fig. 5).

The proposed treatment to the patient was electrocauterization or laser therapy of skin lesions.

### 3. Discussion

Hereditary benign telangiectasia was first described in 1971 by Ryan Wells [15], the condition being present in several members of the same family. Cutaneous lesions usually appear in childhood, but cases with neonatal appearance were described.

Clinically round, oval, dendritic or dotted shape telangiectasias and cutaneous angiomas arranged in plaques or diffuse are present. HBT occurs more often in women, being particularly affected the light-exposed areas such as face, edge of

the lips, neck and upper thorax [10], [20]. Cutaneous lesions are asymptomatic and remain unchanged for a long time without affecting the health status of patients and causing only aesthetic problems [12], [20]. Their appearance may change with time and the aging of the skin can mask telangiectasias. Unlike hereditary hemorrhagic telangiectasia (HHT) in HBT mucous membranes are not affected (without epistaxis or internal bleeding), or affecting other organs such as the lung or liver.

Histology of the HBT shows a normal epidermis and dilated venules especially in subpapillar plexus, but also dilation of capillaries and arterioles.

The etiopathogeny of HBT remains unknown, assuming the involvement of angiogenic factors, hypersensitivity to estrogen or progesterone in affected areas [10], [21], but these theories have not been proven. Genetic analysis conducted in patients with HBT found a dominant autosomal transmission having as a cause the damage of Hc7-M6 loci on chromosome 5q14, this being identical with the locus CMC1 the gene responsible

for capillary malformations [1].

Two cases of HBT without family history were reported in literature [2], [6], [8]. Onishi found arteriovenous malformations in 10 patients with HBT [10]. The association with familial glomerulonephritis has also been described [18].

The differential diagnosis should be done mainly with *HHT (Rendu Osler Weber syndrome)*. HHT is a disease with autosomal dominant transmission. Clinically, HHT presented with telangiectasia as macules and papules, with size of a pinhead to several millimeters, dotted and rarely linear or spider-like [5], [16]. The locations are mostly on the face, lips and oral mucosa, tongue, nasal mucosa, ears, hands, chest and legs [7], [11]. Vascular malformations may be present in the lungs, liver, gastrointestinal tract, genitourinary tract, etc. [7]. This condition is characterized by recurrent epistaxis, with onset prior to 20 years. The bleeding may be also as hematuria, intestinal bleeding, but very rarely as cutaneous haemorrhages.

*Generalized essential telangiectasia* is a rare condition with onset typically in women aged 40-50 years, but with the possibility of onset in childhood or in young adults. Clinically is presented as areas of telangiectasia initially appeared on the lower limbs and slow extension on the trunk and upper limbs which may be accompanied by numbness, tingling or burning sensation in the limbs. Recently, ocular manifestations have been reported as conjunctival telangiectasias [17], [19]. Skin and mucosal hemorrhages are not a characteristic of the disease [4] despite mentioning them by a number of authors [14].

*Unilateral nevoid telangiectasia* may be congenital or acquired, in the last case occurring during puberty or pregnancy, periods of life with high levels of

estrogens. The location of telangiectasias is on the photoexposed areas, with no systemic involvement.

Spider telangiectasia appears as a single or multiple, sometimes unilateral, red papules with radial capillary expansions, usually occurring in pregnancy or liver disease.

*Telangiectasia macularis eruptiva perstans* is a rare form of cutaneous mastocytosis, occurring more frequently in middle-aged adults and rarely in children. Clinically, it present as erythematous or pigmented macules and papules, dotted with telangiectasias, located on the trunk and limbs. [9] Histopathology with toluidine blue stain reveals slightly increased number mast cells situated around the dilated vessels.

*Acquired portwine stain (APWS)* is a rare form of portwine stain, with onset after birth. APWS is observed after local trauma. It may also occur during the pregnancy or the use of contraceptives by increasing estrogen levels. [3] Clinically, APWS presents as erythematous macules with fewer or no telangiectasias on their surface, localized unilateral on the face and neck.

The treatment of HBT usually is not necessary as these are benign lesions. Treatment consists of electrocauterization, laser therapy or their combination. In two separate studies an improvement could be achieved by using the intense pulsed light therapy and pulsed dye laser [8], [13]. Oral tetracycline was effective in treating of primaries teleangiectasias [21].

#### 4. Conclusion

HBT is a benign condition that must be known in the context of conditions associated with telangiectasia.

**References**

1. Brancati, F., Valente, E.M., Tadini, G., Caputo, V., Di Benedetto, A., Gelmetti, C., et al.: *Autosomal dominant hereditary benign telangiectasia maps to the CMC locus for capillary malformation on chromosome 5q14*. In: *J Med Genet* (2003), vol. 40, p. 849-853.
2. Cai, L., Sun, Q.M., Zang, D.J., Zhang, J.Z.: *Hereditary benign telangiectasia without family history in China*. In: *Chin Med J (Engl)*. (2011), vol. 124(5), p. 795-6.
3. Cobb, M.W., Goldman, L.: *Acquired port-wine stain*. In: *J Am Acad Dermatol* (1990), vol. 22, p. 688-690.
4. Dowd, P.M., Champion, R.H. *Disorders of blood vessels*. In: *Textbook of Dermatology*, Champion, R.H., Burton, J.L., Burns, D.A., Breathnach SM(eds). Blackwell Science, Oxford: Vol. 3, 6th ed., (1998), p. 2073-97.
5. Faria, D., Green, H., Fivenson, D.: *Peripheral vascular disease*. In: *Dermatology*, Moschella S, Hurley H, (eds.). WB Saunders, Philadelphia, (1992), p.1145-90.
6. Liuti, F., Almeida, P.J., Borrego, L., Montenegro, T. *Hereditary benign telangiectasia in monozygotic twins with no family history*. In: *Int J Dermatol*. (2014, Dec 29). doi: 10.1111/ijd.12721. [Epub ahead of print]
7. Mad, J.F. *Generalized angiomatosis (telangiectasia)*. In: *JAMA* (1934), vol. 102, p. 442-7.
8. Nakajima, I., Okuyama, R., Terui, T., Tagami, H., Aiba, S. *The first report of non-hereditary benign telangiectasia*. In: *J Eur Acad Dermatol Venereol* (2006), vol. 20, p. 1329-1331.
9. Oanta, A., Irimie, M., Oanta, S., Veress, N. *Telangiectasia macularis eruptiva perstans*. In: *DermatoVenerol. (Buc.)* (2012), vol. 57, p. 151-154.
10. Onishi, Y., Ohara, K., Shikada, Y., Satomi, H. *Image analysis of hitherto unknown association with arteriovenous malformation*. In: *Br J Dermatol* (2001), vol. 145, p. 641-5.
11. Plauchu, H., de Chadarevian, J.P., Bideau, A., Robert, J.M. *Age-related clinical profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population*. In: *Am J Med Genet*. (1989), vol. 32(3), p. 291-7.
12. Puppin, D. Jr., Rybojad, M., Morel, P. *Hereditary benign telangiectasia: two case reports*. In: *J Dermatol* (1992), vol. 19, p. 384-6.
13. Purcell, E., Condom, C. *Intense pulsed light therapy in the management of hereditary benign telangiectasia*. In: *Br J Plast Surg* (2004), vol. 57, p. 453-455.
14. Rothe, M.J., Grant-Kels, J.M. *Nomenclature of the primary telangiectasia*. In: *Int J Dermatol*. (1992), vol. 31(5), p. 320.
15. Ryan, T.J., Wells, R.S. *Hereditary benign telangiectasia*. In: *Trans St Johns Hosp Dermatol Soc* (1971), vol. 57, p. 148-56.
16. Sanchez, J., Ackerman, AB. *Vascular proliferation of skin and subcutaneous fat*. In: *Dermatology in general medicine*, Fitzpatrick TB, Eisem AZ, Wolff K et al. (eds.). McGraw-Hill, New York: (1993), p.1209-43.
17. Seiter, S., Gantenbein, C., Ugurel, S., Ruprecht, K.W., Tilgen, W., Reinhold, U. *An oculocutaneous presentation of essential progressive telangiectasia (Letter)*. In: *Br J Dermatol*. (1990), vol. 140, p. 969-71.

18. Sherwood, M.C., Pincott, J.R., Goodwin, F.J. *Dominantly in herited glomerulonephritis and unusual skin disease*. In: Arch Dis Child (1987), vol. 62, p. 1278-80.
19. Swensson, B., Swensson, O., Haring, G. *Progressive disseminated essential telangiectasia with conjunctival involvement*. In: Klin Monbl Augenheilkd. (1998), vol. 212(2), p. 116-9.
20. Wells, R.S. *Hereditary benign telangiectasia*. In: Br J Dermatol (1971), vol. 84, p. 93-4.
21. Zahorcsek, Z.S., Schneider, I. *Hereditary benign telangiectasia*. In: Dermatology (1994), vol. 189, p. 286-8.