

Reply,

Hereditary benign telangiectasia

Response to "Letter to the editor" by Ozsoylu entitled "Hereditary benign telangiectasia?"

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Ozsoylu commented on our article entitled 'Two cases of hereditary benign telangiectasia in Turkey: sporadic occurrence with punctate telangiectasias surrounded by anemic halos'. He concluded that endoglin gene mutation better be searched to separate these children's telangiectasia from hereditary hemorrhagic telangiectasia.

Hereditary hemorrhagic telangiectasia (HHT, or Osler-Rendu-Weber syndrome) is an autosomal dominant disease characterized by an aberrant vascular development that results in a range of vascular malformations from smaller mucocutaneous telangiectasias to large visceral arteriovenous malformations. The clinical manifestations of HHT include recurrent epistaxis, multiple telangiectasias at characteristic sites (lips, oral cavity, nose, fingers), visceral lesions, such as gastrointestinal telangiectasias, pulmonary, cerebral or hepatic arteriovenous malformations¹.

Curaçao's diagnostic criteria for HHT includes the following four criteria; spontaneous recurrent epistaxis, multiple telangiectasias at characteristic sites, family history and visceral involvement. Three criteria must be set for the definite diagnosis of HHT². However, we must keep in mind that these diagnostic criteria were installed for adults and can be misleading when applied to children. Symptoms and signs of HHT generally develop during childhood and adolescence, such that the absence of epistaxis, telangiectases or symptoms of solid organ AVM is common in affected children. Children with a family history should undergo molecular genetic testing to approve or reject the diagnosis. Two different mutations in the gene's encoding endoglin (ENG, chromosome 9q34) and activin A receptor type-like kinase 1 (ALK-1, chromosome 12q13) have been described resulting in HHT types 1 and 2, respectively. And these clinical criteria are likely to be further refined as molecular diagnostic tests become available in the subsequent years^{2,3}.

We presented two 13 and 16 years old girls with telangiectasia on the outer side of the labial mucosa and telangiectasias on sun exposed areas like face and back of the hands and forearms. Both of the patients were otherwise in good health, with no hemorrhagic episodes or systemic disorders. None of the both family members had any history of similar lesions or hemorrhagic episodes. When the patients undergone a full physical examination any telangiectasia or haematic dots were not detected in the mucosal surfaces of ophthalmic and ear-nose-oral-throat. Hematological and biochemical analyses were unremarkable. Abdominal ultrasonographic evaluation revealed no hepatic vascular malformation. On the basis of these findings, a clinical diagnosis of hereditary benign telangiectasia was made. Genetic testing was not performed because of the patients' history, clinical findings and facility. But as Ozsoylu accentuated, analysis of gene mutation would be better to separate these children's telangiectasia from hereditary hemorrhagic telangiectasia and confirm or refute the molecular diagnosis in the patients.

REFERENCES

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