

Suntharesan J¹, Perera TMR¹, Vipulaguna VD¹, Wijesekara DS¹, Wettasinghe CA², Kumarasiri MKHN², Niwanthika TKI², Siriwardana HD²

¹University Paediatric Unit, Colombo South Teaching Hospital

²Department of Paediatrics, University of Sri Jayewardenepura

Background - Hereditary haemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu syndrome, a rare autosomal dominant disorder that affects blood vessels throughout the body and results in bleeding tendency. Forms of HHT include types 1, 2, 3 and juvenile polyposis/HHT syndrome. Its incidence is 1 in 5,000 to 10,000.

Method - A 12-year-old boy who was investigated for epistaxis for 2 years of age, presented with worsening of epistaxis, throbbing type frontal headache over six months with a progressively increasing vascular birth mark over right upper limb. His mother and sister also had spontaneous epistaxis. Examination revealed a pinkish purple papular vascular malformation on medial side of upper limb with superficial telangiectatic vessels. There was telangiectasia on trunk, limbs and face. Rest of the examination was normal.

Results - Basic hematology, chest X-ray and 2D echo were normal. Contrast CT brain and MRI brain revealed L/middle cerebral artery vascular malformation. Four vessel carotid angiogram revealed large L/parietal carotid AV fistula between L/middle cerebral artery and cortical veins with variceal enlargement.

Child underwent open craniotomy and resection of AV fistula and had uneventful recovery.

Conclusion - Diagnosis was made clinically on the basis of Curacao-criteria, established by the Scientific Advisory Board of the HHT Foundation, which are epistaxis, telangiectasia, visceral arterio-venous malformations and a first-degree relative who has HHT.

Our case fulfilled the Curacao criteria. Current recommended treatment for arterio-venous Malformation (AVM) depends on its size and location. Interventional Radiologists offer non-surgical treatment for AVM. Invasive treatment of brain AVMs include endovascular embolization, surgical resection, and focal beam radiation alone or in combination.