A Truly Unusual Overgrowth Syndrome: An Alternative Diagnosis to Klippel-Trénaunay-Weber Syndrome

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To the Editor I read with interest the case report by Yazaki et al (1) in which an adult patient was given the diagnosis of Klippel-Trénaunay-Weber syndrome based on the presence of hemitruncal port-wine stain, subcutaneous AV fistulae and lower extremity varicose veins. Several points regarding this diagnosis warrant comment.

First, Klippel-Trenaunay-Weber syndrome is a confusing and inaccurate designation. Using this eponym “Klippel-Trenaunay-Weber” precludes determination of whether a patient has Klippel-Trenaunay syndrome (KTS) or Parkes Weber syndrome (PWS) (2). These two overgrowth syndromes, while they may share some of the clinical features, represent separate clinical entities with different pathogenesis and natural history. Unfortunately, many inaccurate eponyms are still commonly used in the vascular anomalies literature.

Second, the key vascular components of KTS are capillary-lymphatico-venous malformation, hence the name CLVM. Arteriovenous malformations, as seen in this patient, are not part of the clinical manifestation of KTS. The association of port wine stains and high flow shunts can be seen in some rare syndromes such as Parkes Weber syndrome, capillary malformation-arteriovenous malformation syndrome (CM-AVM; RASA1) mutation (3), Cobb syndrome and CLOVES syndromes (4, 5). The CLOVES syndromes is a recently described phenotype that associates congenital lipomatous overgrowth, vascular malformations, epidermal nevi and spinal/scoliosis/seizures/skeletal anomalies (4-6). One of the common vascular findings in this syndrome is a paraspinal-truncal arteriovenous shunt associated with a port wine stain and truncal overgrowth. Cobb syndrome is a metameric high flow shunt that involves an entire somatic block; lack of spinal involvement in the present patient makes this diagnosis unlikely.

Third, hereditary hemorrhagic telangiectasia (HHT) is also unlikely to be the proper diagnosis; as the major cutaneous birthmark shown in the photographs (Fig. 1 in the case report) represents a large capillary malformation, not the classic mucocutaneous telangiectasia seen in HHT. In addition, the absence of cardinal signs of HHT, such as epistaxis and family history, among others, further excludes HHT from the differential diagnosis.

Reviewing another presumed case report of KTS and hepatic shunt cited by the authors, Yamagami et al focused on the technical aspect of the embolization of an pediatric Ab- ernethy type 2 shunt and failed to mention any of the classic features of KTS in their patient (7). This was a congenital hepatic shunt in a child. In contrast, Yazaki et al (1) described an adult intrahepatic shunt (which is typically an acquired lesion). These two types of shunts are etiologically and prognostically different.

Finally, and despite the continuous advancement of our knowledge in the field of overgrowth syndromes with complex vascular anomalies, misdiagnosis is still common (8). Clinical overlap between these phenotypes often makes the diagnosis difficult as many patients with overgrowth syndromes do not belong to any recognizable condition (9).

I believe that the case presented here does not belong to KTS. Therefore, I suggest further analysis of the clinical manifestation in this unusual case to reach the appropriate diagnosis.

Ahmad I. Alomari

References

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