Case 5

Extensive reticulate capillary malformation since birth

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Patient: A Thai male newborn from Bangkok

Chief complaint: Extensive reticulate capillary malformation and right leg hypertrophy since birth

Present illness:

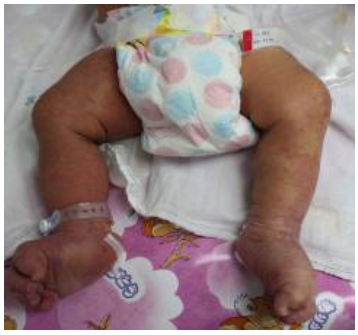
A Thai full-term male newborn was born to a nonconsanguinous couple by cesarean section with Apgar score of 9 and 10 from Klang hospital. His mother was a healthy 20-year-old woman with no perinatal problem. Shortly after birth he was found to have extensive pink to erythematous patches and slightly enlarged of Right leg. Then he was refered to Ramathibodi hospital for concerning of complex-combine vascular malformation syndrome.

Past history:

- He was the first child of his mother.
- No family history of vascular anomaly and limb overgrowth.

Skin examination:

- His head circumference was 35 cm. (within normal range)
- Extensive reticulate pink-red continuous patches involving right torso, right arm, both legs and nearly entire back with midline demarcation was noticed on anterior trunk.
- Right leg was obviously larger than Left side.
- Increase skin markings on both soles was detected.



Diagnosis: **Diffuse capillary malformation with overgrowth**

Investigation:

Ultrasound abdomen: No abdominal mass, no hepatosplenomegaly.

Doppler ultrasound both legs:

- No vascular or lymphatic malformation was detected.

Treatment: Follow up and family counselling

Discussion:

Vascular anomalie classification was accepted at the 1996 biennial meeting of the International Society for the Study of Vascular Anomalies (ISSVA).[1,2] Vascular anomalies were divided into two main categories: vascular tumors, produced by cell proliferation, and vascular malformations, characterized by abnormal distorted vascular channels.

For vascular malformations, are divided into two categories based on flow characteristics and channel morphology. Fast-flow vascular malformations consist of arterial malformations, arteriovenous fistulae (AVFs) or arterial venous malformations (AVMs). Slow-flow vascular malformations include venous, lymphatic and capillary malformations. In addition, there are also various types of complex and combined vascular malformations or it can be part of a syndrome.

By the way, up until now there is no report of vascular tumor associated with limb overgrowth.

Overgrowth of the limb with the coexistence of a capillary malformation, as in our patient, must be differentiated from Parkes Weber syndrome, diffuse capillary malformation with overgrowth (DCMO), Klippel-Trenaunay syndrome (KTS) and Proteus syndrome.[3]

1) Parkes Weber syndrome was described in the early 1900s. Combined vascular malformation very similar to KTS, is clinically and it can be differentiated from KTS by the presence of AVFs. Clinical findings may include the following: a congenital cutaneous red stain, dilated veins (often with a thrill), cutaneous warmth, pulsating lesions, audible bruit, an enlarged extremity with excess length progressively developing until the end of puberty

2) Diffuse capillary malformation involving an entire limb with congenital hypertrophy of the limb, there is no further progression of the overgrowth after birth. The entire length of skin covering the limb reviews diffuse capillary malformation, often involving a much wider area of skin. No varicose veins and/ or venous malformation, as seen in KTS, is detected.

3) Klippel-Trenaunay syndrome (KTS) consists of classic tirad of capillary malformation of the affected extremity, underlying bony and soft tissue hypertrophy, and varicose veins and/ or venous malformation. Care must be taken with young children, because varicosities or VMs are reported to be present in around 72% of KTS patients, with an incidence of less than 60% in patients under 5 years of age; this prevalence increases with age[4], As for this reason, color Doppler analysis should not be performed too early in life because it usually results in conflicting results.

4) Proteus syndrome is a somatic activating mutation *AKT1* gene.[5] Manifestations include a cerebriform connective tissue nevus; epidermal nevi; vascular malformations; capillary malformation; dysregulated adipose tissue; pulmonary abnormalities; disproportionate, asymmetric overgrowth with skeletal defects; and a distinct facial phenotype. However, children with later diagnosed Proteus syndrome were usually normal at birth or had mild-to-moderate alterations, hyperplasias, hamartomas, or vascular malformations.[6]

For our patient, a male newborn with extensive reituculate capillary malformation and Right leg hypertrophy since birth. Parkes Weber syndrome can be completely ruled out due to Doppler ultrasound result. According to morphology, capillary malformation lesions and sharp midline demarcation as described in DCMO, currently we prefer this spectrum of diasease for our patient.

DCMO was first proposed by Margaret S. Lee et al.[7] In 2013. Typical presentation include, reticulate, pale, extensive, and diffuse, in multiple anatomic regions that are stained contiguously. And soft tissue and/or bony overgrowth that is proportionate to that of the child. For DCMO, Margaret S. Lee et al identified 73 patients without major complications during follow-up and the maximum period of follow up for any individual patient was 14 years.

As previously discuss, KTS and proteus syndrome are not obviously present fully at birth. Therefore, our patient required periodic follow-up to monitor for it.

References:

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