Parkes Weber Syndrome Involving Right Lower Limb: A Case Report

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Abstract Vascular malformations (VMs) are developmental abnormalities of the vascular system. Malformations may involve any segment of the vascular tree: arteries, capillaries, veins or lymphatics. High-flow arteriovenous malformations (AVMs) are associated with shunting of large amounts of arterial blood into the venous system; these lesions can have dynamic and dramatic hemodynamic manifestations, such as massive arteriolisation with gross venous engorgement, organomegaly of concerned anatomical region and high-output cardiac failure. Patients with Parkes Weber syndrome have clinically significant micro- or macrofistulous arteriovenous shunts, affecting usually one extremity. The patient has dilated, frequently visible pulsatile varicose veins and other visible signs of AV shunting. The abnormality is sporadic; it is likely a somatic mutation. There is frequent intraosseous involvement. The presence of high AV shunts differentiates Parkes Weber syndrome from Klippel–Trenaunay syndrome which is a clinical triad of capillary malformation, soft tissue and bone hypertrophy and venous and lymphatic malformations. The mutations in RASA 1 gene leads to Parkes Weber syndrome characterised by the presence of multiple, small (1–2 cm in diameter) capillary malformations mostly localized on the face and limbs. We report a case of congenital AVM

of the lower limb causing cardiac decompensation. The patient was safely and successfully treated by performing a knee disarticulation.

Keywords Vascular malformations · Arteriovenous shunts · Massive arteriolisation · Cardiomegaly · Parkes Weber syndrome

Introduction

Vascular malformations (VMs) are developmental abnormalities of the vascular system. The mutations in RASA 1 gene leads to Parkes Weber syndrome characterised by the presence of multiple, small (1–2 cm in diameter) capillary malformations mostly localized on the face and limbs [1].

Case Report Article

A 32-year-old female patient came with complaints of torrentially bleeding ulcer on the right forefoot since 5 days, which stops on pressure on the bleeding ulcer and elevation of the leg. From birth to 6 years of age, there were no signs and symptoms. At 6 years, she noticed increased in size of the right leg as compared to the left. Doppler study of the right lower limb showed abnormal vasculature and no active management. The discrepancy between the two lower limbs went on increasing to the age of 12 years without any other symptom, which was treated by specialised shoes. She got married at the age of 22 years, during the second pregnancy, the right leg size increased

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Fig. 1 Multiple pulsatile varicose veins in right lower limb

exponentially and she developed an ulcer near the right little toe which was not bleeding. The limb size decreased and the ulcer healed after the second delivery. After 2 years, she developed an ulcer over the right forefoot which used to bleed torrentially for which she underwent angiography showing AV malformations in the foot. Again, she was advised conservative treatment. Since the past 3 years, she was not using her right lower limb for the fear of bleeding.

General examination: A thin built lady with severe pallor, pulse rate 130/min, water hammer pulse with machinery murmur was discernible. BP readings in supine position.

150/0 mmHg—right arm, 150/0 mmHg—left arm
160/0 mmHg—left leg, 160/0 mmHg—right leg

Local examination: A 3×2 cm oval-shaped ulcer is present over the dorsum of the right foot, with continuous spurting of arterial blood. The right lower limb has multiple dilated, tortuous veins from mid-thigh more on medial aspect of knee, to the dorsum of the foot which are pulsatile (as shown in Fig. 1).

Length of extremities

Lt lower limb—116 cm

Rt lower limb—120 cm

Investigations: Chest x-ray shows cardiomegaly (as shown in Fig. 2).

2D Echo; EF—70 %, dilated left and hypercontractile left ventricle. Angiography was done in January 2013 which showed hypertrophied right profunda femoris and its branches, right superficial femoral artery, right popliteal artery with normal trifurcation, diffuse arteriovenous malformations noted involving right leg and foot supplied by anterior and posterior tibial and peroneal arteries consistent with diffuse complex AV malformations involving right leg and foot supplied by anterior, posterior tibial arteries and peroneal arteries for which no endovascular management is feasible. Digital Subtraction Angiography was done on March 2013, suggesting AV fistula present below level of malleolus of the right foot “as shown in Fig. 3.” All other arteries are hypertrophied and dilated.

Fig. 2 Preoperative chest X ray showing cardiomegaly

Fig. 3 DSA scan showing arteriovenous fistula below level of malleolus of right foot

Fig. 4 DSA scan showing aneurysm of profunda femoris and multiple aneurysms of deep popliteal and femoral artery

Fig. 5 Intraoperative photograph showing tibial and femoral condyles

Fig. 7 Abnormal vascularity over periosteum of femoral condyles

Aneurysm of profunda femoris and multiple aneurysms of deep popliteal and femoral artery “as shown in Fig. 4.” The case was diagnosed as Parkes Weber Syndrome.

Treatment

Medical management: To decrease the preload, IV labetalol 1 mg/min in infusion pump was given.

Surgical management: by keeping in mind the opinion of interventional radiologist and vascular surgeons, collective decision was taken to disarticulate the right

knee joint “as shown in Figs. 5 and 6.” We intentionally avoided amputation through bone to avoid the risk of uncontrollable bleeding from the bone end (as shown in Fig. 7).

Discussion

Management of arteriovenous malformation (AVM) varies, with a conservative approach adopted for patients that are asymptomatic or have minor symptoms. If treatment is required, techniques that may be used include catheter embolization or direct percutaneous sclerotherapy [2]. Multidisciplinary approach is central to the effective management of AVM. Despite multiple procedures, total eradication is often not achieved. For some patients, limb amputation becomes necessary and carries significant risk. Cardiac decompensation is an absolute indication for surgery [3] “as shown in Fig. 8.” Schwartz and associates reported on 82 patients treated at the Mayo clinic for AVM; 18 patients required amputation at various levels of extremity. Still, for many patients, amputation means cure and a prosthesis offers the possibility of functional recovery [4]. In our patient, repeat embolisation would not have been appropriate as it was attempted but failed due to the diffuse nature of the AVM with multiple nidi and hyperdynamic circulation. Our patient had a life threatening, symptomatic, nonfunctional lower limb AVM. In this situation, we believe that disarticulation should be explained at the patient outset. In the year

Fig. 6 Ligation of popliteal vessels

Fig. 8 Postoperative chest X-ray showing decreased cardiomegaly

2010, a case of upper limb disarticulation for a patient of congenital AV malformation has been reported in China till date [5].

Conclusion

Arterio - Venous Malformations are high-flow lesions having a direct connection between an artery and vein which remains a surgical challenge. Those affecting the lower limb causing cardiac decompensation are rare. Despite multiple procedures, total eradication is often not achieved. For some patients, the affected limb becomes life threatening in which limb disarticulation/amputation becomes a life-saving surgery.

References

1. Bayrak-Toydemir P, Stevenson D. RASA1-Related Disorders (2011) In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK52764/>
2. L T Cotton, B J Sykes (1956) The Treatment of Diffuse Congenital Arteriovenous Fistulae of the Leg. Ann R Coll Surg Engl 18(2):73-98
3. Phuong D, Nguyen Oriana D, Cohen Evan P (2009) Nadler Arteriovenous malformations, text book of surgery. Chapter no 112 18
4. Peter gloviczki et al (2005) vascular malformations, chapter no 11, volume no 10, page 183–198
5. Yi-Hsuan Su, MD, Vasu Karri, BSc(Hons), MRCS, MSc, Lai-Fan Lu, MD, and Hung-ChiChen, MD, FACS (2010) Upper-Limb Disarticulation for Life-Threatening High-Flow Arteriovenous Malformation: Report of a Case. Vol. 198, no 8