CLINICAL QUIZ (p258-259) ANSWER

Figure 1 reveals cardiomegaly and plethoric lung fields in keeping with heart failure. Figures 2a to 2e show a mid-line round mass within the pineal region; T2-weighted magnetic resonance imaging (MRI) (Figure 2b) shows large signal voids caused by high flow in the vein of Galen, falcine sinus and the confluence of sinuses; magnetic resonance arteriogram (MRA) (Figure 2d) shows the dilated aneurysmal sac with numerous choriodal arteries supplying the confluence which drains into the straight sinus. A diagnosis of a choroidal type of vein of Galen malformation (VGM) is thus made. The girl underwent embolization on day 4 of life. However, she developed sudden clinical deterioration with multi-organ failure in the subsequent month, and was electively extubated on day 43 of life due to her poor prognosis. Skin fibroblasts were saved postmortem for genetic analysis.

The precursor of vein of Galen, or the great cerebral vein, is the large median prosencephalic vein of Markowski. In patients with VGM, arteriovenous shunts, with their arterial supply largely derived from choroidal arteries, develop during the 6th to 11th weeks of gestation. The high blood flow causes the anterior segment of the median prosencephalic vein of Markowski, which normally regresses, to instead progressively enlarge and form the aneurysmal component.[1]

VGM can be classified into the choroidal or mural type according to the angioarchitecture. The former, encountered in most neonates and with more severe symptoms, involves the interposition of an extensive arterial network of several choroidal arteries; the latter is characterised by direct arteriovenous fistulas within the wall of the median prosencephalic vein of Markowski. VGM should be differentiated from vein of Galen aneurysmal dilatation (VGAD), which represents a dilated but normally formed vein of Galen caused by an adjacent brain arteriovenous malformation.

The aetiological basis of VGM is largely unknown, although an association with capillary malformation-arteriovenous malformation (CM-AVM), a newly recognised autosomal dominant disorder caused by mutations in the *RASA1* gene, has been reported.[2] Mutations in *RASA1* gene, which is located on the long arm of chromosome 5, prevent the production of functional p120-RasGAP protein which normally controls RAS/MAPK signaling and lead to the vascular abnormalities seen in people with CM-AVM, including VGM, and Parkes Weber syndrome. In our patient, no mutation was detected in the coding part of *RASA1* gene by sequencing.

Clinical pictures are correlated with the age of presentation and the underlying pathophysiology. Feeding difficulties, tachycardia, plethoric lung fields together with echocardiogram findings in our patient pointed towards high-output heart failure, a common presentation in symptomatic neonates. Cerebral low-resistance arteriovenous shunts cause increased venous return to the right atrium, followed by pulmonary hypertension, and ultimately congestive heart failure due to increased preload.[1] Infants usually present with hydrocephalus. Older children frequently present with headache and signs and symptoms of subarachnoid haemorrhage.

The presence of patent ductus arteriosus may be beneficial to the haemodynamics status, as in our patient, because it may provide a 'blow-off' channel to the pressure-loaded right ventricle, thus preventing further ventricular dilatation and dysfunction.[3]

The differential diagnoses for a newborn child with gross cardiomegaly, severe right ventricular dilation and congestive heart failure include but are not limited to cardiac causes such as coarctation of the aorta and interrupted aortic arch.[4] When the cardiac failure is unexplained by a cardiothoracic examination, cranial arteriovenous malformations should be suspected, and the characteristic bruit over the fontanelle should be sought for; head ultrasound and diagnostic angiography should be performed.

Imaging is important in evaluating the angioarchitecture, size, location and multiplicity of arteriovenous malformations; in diagnosis, guiding therapeutic decisions and long term monitoring. The persistent limbic arch, as well as persistent venous embryonic routes, such as dorsal diencephalic vein (the so-called epsilon) and the falcine sinus are characteristics of a true VGM. Post-processed time of flight MRA and contrast-enhanced magnetic resonance venogram images in our patient (Figures 3a and 3b) show a network of arterial feeders draining into the dilated venous pouch, keeping with the diagnosis of the choroidal type of VGM.

Routine antenatal ultrasound and fetal cardiovascular assessment have enabled the detection of VGMs in the third trimester of pregnancy.[5] In the postnatal period, echocardiography can demonstrate the haemodynamic changes, while computed tomography (CT) can show a multilobulated, intensely enhancing lesion and ventricular dilation.[1] MRI can identify the presence of venous pouch, fistula, nidus, venous drainage, venous thrombosis and sources of arterial feeders; it is the modality of choice to evaluate the ventricular system and cerebral parenchymal damage. Angiography is the gold standard for the evaluation of VGM, as the anatomy of arterial feeders and the haemodynamics of venous drainage can be precisely demonstrated.

While the past neonatal mortality rate was close to 100%, it has dropped to 52% in a large-scale study in 2006.[6,7] Embolization is the first-line treatment, with the goal of decreasing flow in the VGM and reducing cardiac failure.

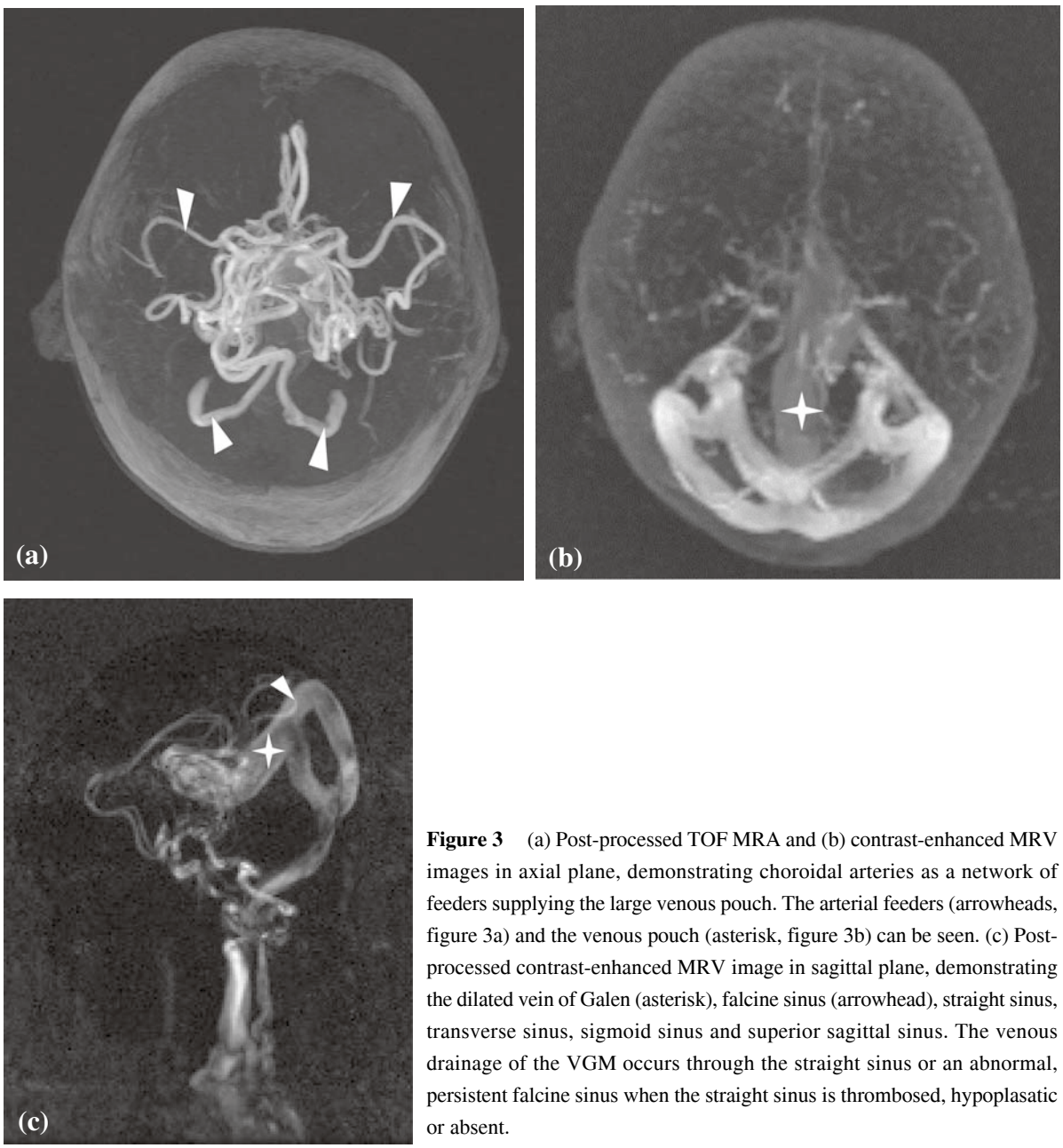

Figure 3 (a) Post-processed TOF MRA and (b) contrast-enhanced MRV images in axial plane, demonstrating choroidal arteries as a network of feeders supplying the large venous pouch. The arterial feeders (arrowheads, figure 3a) and the venous pouch (asterisk, figure 3b) can be seen. (c) Post-processed contrast-enhanced MRV image in sagittal plane, demonstrating the dilated vein of Galen (asterisk), falcine sinus (arrowhead), straight sinus, transverse sinus, sigmoid sinus and superior sagittal sinus. The venous drainage of the VGM occurs through the straight sinus or an abnormal, persistent falcine sinus when the straight sinus is thrombosed, hypoplasatic or absent.

Multiple successive procedures may be required, with embolic glue or detachable microcoils frequently used for occlusion. Our patient received partial embolization via venous access on day 4 of life; more than 30 Guglielmi detachable coils were used. Pre- and post-embolization venograms were shown in figures 4a through 4d.

Medical treatments including diuretics, inotropes and other cardiovascular agents are used to relieve cardiovascular and renal symptoms before surgical embolization.[1] Together with endovascular therapy and a comprehensive multidisciplinary approach, prognosis has been substantially improved in patients with VGM.[8]

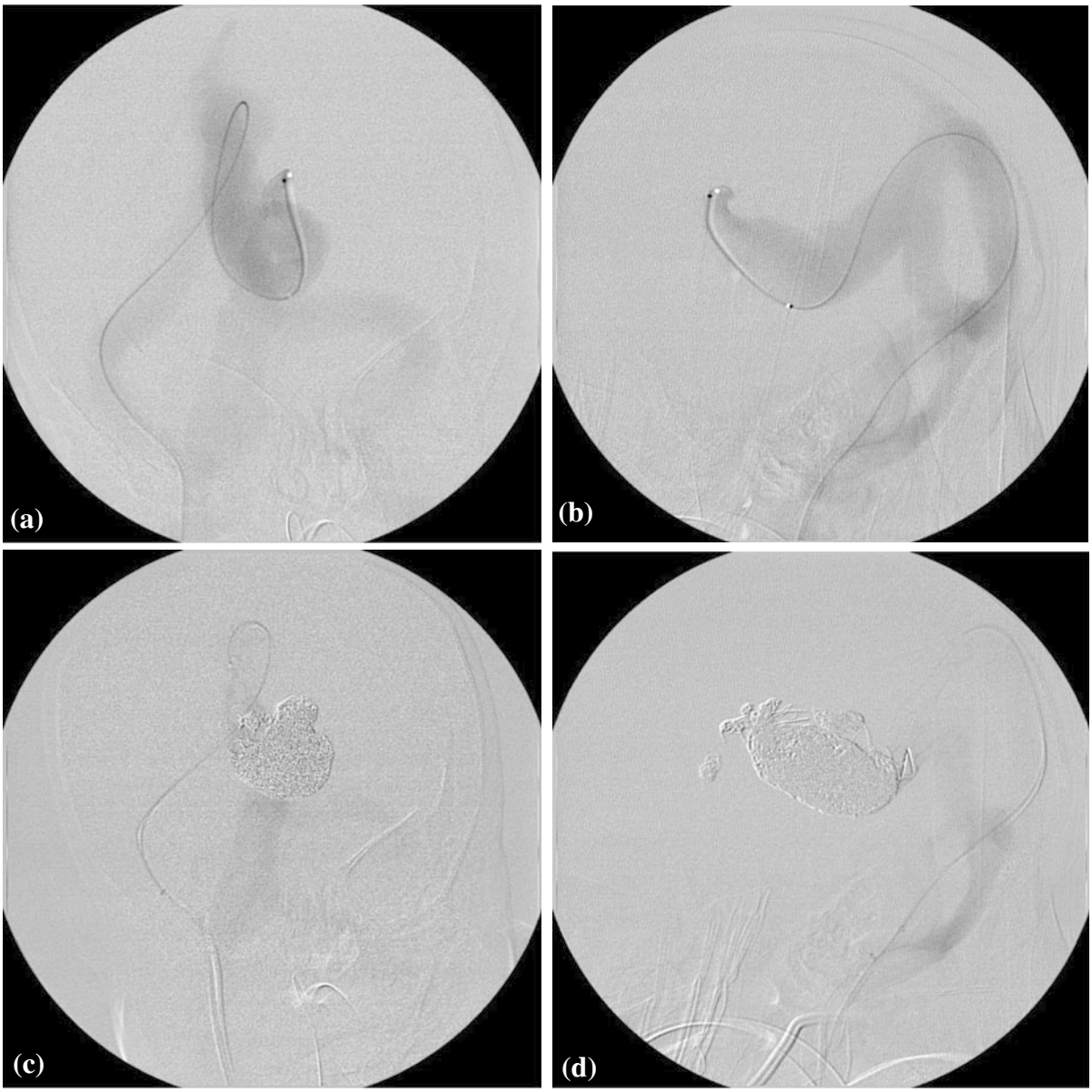

Figure 4 (a) Pre-embolization transvenous digital subtraction angiogram (DSA) in frontal view. (b) Pre-embolization transvenous DSA in lateral view. (c) Post-embolization transvenous DSA in frontal view. (d) Post-embolization transvenous DSA in lateral view.

Figures 4c and d. The venous pouch of the vein of Galen aneurysm and the larger arterial feeders are packed with GDC coils.

References

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