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Case report

Familial incidence of the congenital torcular dural arteriovenous shunt: Case report and review of the literature

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ABSTRACT

Congenital dural sinus malformations are rare but can be major causes of mortality and morbidity in the pediatric population if not detected and managed urgently. Lesions involving large draining sinus structures such as superior sagittal sinus and torcular herophili can result in significant intracranial circulation impairment mostly due to venous drainage disturbance. Early detection plays a pivotal role in the outcome of the patients. Rarely familial incidence of some types of arteriovenous malformations in isolation from other congenital hereditary disorders has been reported. Knowledge of the familial association of congenital dural sinus malformations may raise the awareness for considering the possibility of occurrence of these lesions in the relatives of index cases. Herein, we describe the occurrence of giant torcular dural shunt in two pediatric cousins treated with endovascular embolization.

1. Introduction

Several hereditary syndromes have known associations with CNS arteriovenous malformations (AVMs) including Sturge-Weber, Klippel-Trenaunay-Weber, Parkes-Weber, hereditary hemorrhagic telangiectasia (HHT), and hereditary neurocutaneous angiomatosis. Furthermore, the familial incidence of AVMs in isolation from other congenital hereditary disorders has also been reported. Congenital dural sinus malformations (DSMs) are rare but clinically remarkable particularly in the pediatric population. Herein, we describe the occurrence of giant torcular dural arteriovenous shunt (AVS) in two pediatric cousins treated with endovascular embolization.

2. Case presentation

2.1. Case 1

A 5 month old male presented with the chief complaint of poor head control, right head tilt, and increasing head circumference with full and pulsatile fontanelle. Imaging studies revealed marked venous dilation adjacent to the vein of Galen. He underwent multiple sessions of embolization and radiation therapy without significant improvement (Fig. 1). Ten months later the patient presented to our center with developing seizure and apnea requiring immediate intubation. Immediate cerebral angiography demonstrated the filling of the large cavity within the mostly thrombosed torcular with retrograde drainage into the superior sagittal sinus (SSS) and subsequently into the deep cerebral vein via a second channel. Embolization of the torcular and the distal third of the SSS was performed utilizing multiple hydro coils (Cosmos, Hydrocoil, Microvention, Tustin, CA), pushable fiber coils (Nester, Cook, Bloomington, IN), *N*-butyl cyanoacrylate (NBCA, Codman, Raynham, Massachusetts), and ethylene vinyl alcohol copolymer (Onyx, ev3, Irvine, CA). Further endovascular management included stenting of the very stenotic distal left sigmoid sinus. Although there was minimal residual arteriovenous shunting from right external

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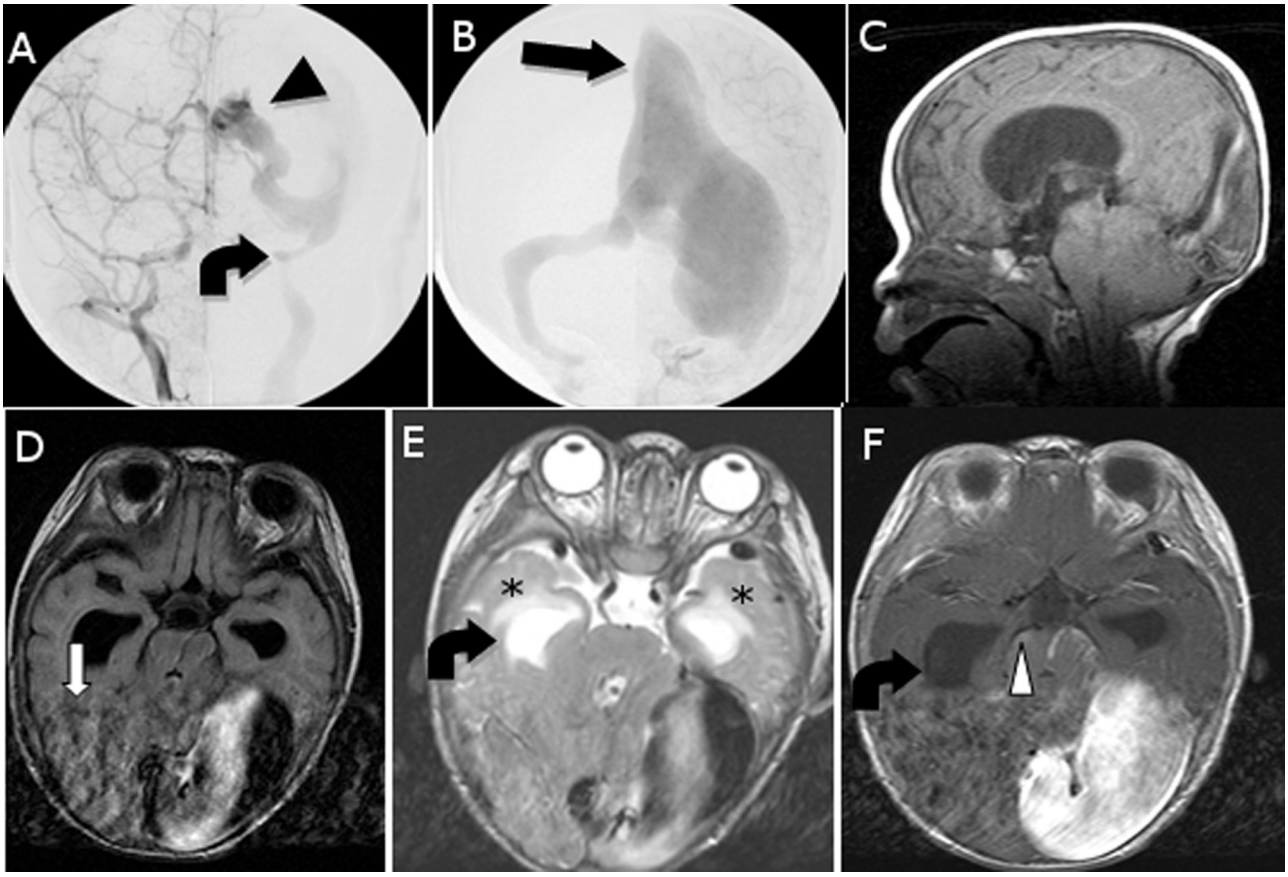

Fig. 1. (Case 1): (A, B): Images from an angiogram performed at an outside institution, demonstrating AV shunting from ECA branches into a large recipient venous pouch (arrowhead) secondarily draining into the massively dilated left transverse, sigmoid and distal SSS (arrow). A focal stenosis of the sigmoid sinus at the junction of the internal jugular vein is also observable (curved arrow). (C–F): MRI brain T1W sagittal (C), T1W axial (D), T2W axial (E), and axial T1 post-gadulinium (F) images demonstrate mass effect and downward displacement of the cerebellar tonsils as a consequence of venous hypertension. Significant phase (pulsation) artifact (D, open arrow) indicates the large amount of flow and pulsation within the dural sinus malformation. Hydrocephalus (curved arrows) and transependymal CSF flow (asterisk) is evident as a consequence of venous hypertension. Abnormal dilation of paranchymal veins (venous collateral pathways) of the posterior fossa structures is observable (F, arrowhead).

carotid artery branches to the posterior wall of the torcular, significant improvement in arteriovenous shunting was achieved.

Multiple annual follow-up angiographies revealed significant improvement in the intracranial circulation and venous hypertension without any evidence for recurrence of the high-flow torcular shunt; however, a small residual low flow arteriovenous fistula (AVF) between branches of the left superior cerebellar artery and the left pica draining into the dural veins without extending into the pial/cortical venous system remained. Although patient achieved significant improvement in clinical outcome, prolonged neurological sequels due to delayed diagnosis remained. Follow-up visits were negative for any coagulopathy or other relevant signs and symptoms such as skin lesions.

2.2. Case 2

The second case was the third-degree relative (paternal cousin) of case 1. A 20 day-old female was admitted for evaluation of failure to thrive, dysphagia, and enlarged head circumference with bulging fontanelles. Pregnancy history was significant for intrauterine diagnosis of an intracranial mass superior to the posterior fossa which was identified on 20 week gestation ultrasound. Amniocentesis with chromosomes and single nucleotide polymorphism array examinations were reported normal. Mother denied smoking and drinking alcohol during pregnancy and her past medical history was significant for one experience of miscarriage. Family history was negative for hematological disorders, cardiovascular events, stroke prior to age 50, and frequent miscarriages.

Fetal MRI revealed a midline mass in the region of the torcular with signal characteristics of blood or blood products that was thought to be consistent with dural sinus thrombosis (Fig. 2A, B). Serial ultrasounds were performed which demonstrated some initial growth of the mass, but subsequently the lesion size stabilized. Following delivery, a CBC was obtained as part of a sepsis work-up and thrombocytopenia was detected (53000/μL) which was improved gradually suggestive for being consumptive in nature due to the intracranial thrombosis. Neurological, cardiovascular, and neonatal physical examinations were normal and no coagulopathy and skin lesions were detected. She remained clinically stable and was discharged on day 6 following caesarian section. Further diagnostic investigation revealed large secundum atrial septal defect, mild to moderate pulmonary artery stenosis, anomalous aortic origin, and coronary artery anomaly. Subsequent genetic screening was positive for a heterozygous missense mutation of the PTPN11 gene which was consistent with the clinical diagnosis of Noonan Syndrome.

MRI revealed posterior extra-axial heterogeneous signal changes at the level of the torcular consistent with the previously described thrombosis/hematoma resulting in significant mass effect on the cerebellum, fourth ventricle, and right occipital hemispheres. (Fig. 2C–G).

Cerebral angiography demonstrated a significant AVS from multiple meningeal vessels including bilateral occipital, middle