International Journal of
Neurological Disorders & Interventions
Case Report Open Access

Foetal Thrombosis of Torcular Herophili: Report of Two Cases and Review of the Literature

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Abstract

Background: Foetal thrombosis of the torcular herophili is a rare entity, usually detected during the 2nd trimester. Prognosis and risk factors for severe outcome are yet poorly characterized, but important for counselling and management.

Aim: To describe two new cases of foetal thrombosis of the torcular herophili and review the literature, focusing on prognosis and predictive factors.

Methods and Results: We retrospectively reviewed clinical data, antenatal and postnatal MRI findings of two cases of foetal torcular herophili thrombosis diagnosed prenatally. Foetal MRI was performed at 36 and 32 weeks' GA, after abnormal ultrasounds at 32 and 31+5 weeks' GA, respectively. Last clinical and MRI postnatal follow-up was performed at 5 years and 15 months, respectively. Both patients had ischaemic brain lesions and neurological impairment. We found 66 cases reported in the literature. The overall mortality rate was 15% while a favorable outcome was seen in 87% of surviving patients. Risk factors for neurological disability were male sex and ischemic brain lesions, while dural arteriovenous shunt or signs of foetal decompensation were associated with foetal demise or post-natal death.

Conclusion: Posterior fossa foetal ultrasound allow suspicion of foetal dural sinus thrombosis, which is confirmed by foetal MRI. Outcome ranges from foetal demise to complete spontaneous regression with normal development. Identified foetal neurological risk factors will help the management of this rare and potentially life threatening condition

Publication History:

Received: May 23, 2017
Accepted: December 27, 2017
Published: December 29, 2017

Keywords:

Dural sinus thrombosis, Dural sinus malformations, Magnetic Resonance Imaging, Foetal, Neonates, Ultrasound.

Introduction

Foetal dural sinus thrombosis (DST) is a rare entity, that is usually initially suspected at the second trimester by routine foetal ultrasound examination (US)[1,2]. Its appearance on ultrasounds may be nonspecific and may lead to misdiagnosis. A prompt referral for foetal magnetic resonance imaging (MRI) confirm the diagnosis and inform on parenchymal involvement [3]. The exact underlying cause of DST is unknown, but most reported cases involve the torcular Herophili (TH) and are associated with a particular type of midline dural sinus malformation (DSM) with giant lakes and low-flow arteriovenous shunts [3-5]. Less than seventy cases of foetal DST are reported in the literature and outcome ranges from foetal demise to complete thrombus regression with normal development. A better understanding of this uncommon condition and of neurological risk factors is warranted for a timely diagnosis and a more precise definition of its prognosis. Besides, early diagnosis may allow for proper prenatal counselling and an adequate planning of postnatal management.

We report two cases of foetal torcular DST (tDST) diagnosed at the third trimester and we review the pertinent literature with focus on risk factors for severe outcome.

Methods

The radiological findings and clinical features of two patients diagnosed at the University Hospital of Padua were retrospectively reviewed. The foetal MRI was performed on a 1.5-T MR scanner (Achieva, Philips, Best, The Netherland). A postnatal 5-year and 15-month follow-up was performed by a paediatric neurologist in patients 1 and 2, respectively.

The literature review included an extensive search in Pubmed for case reports of foetal DST.

The literature search was conducted through PUBMED, using the following search term combinations without additional filters: "fetus" [All Fields] AND "cerebral thrombosis" [All Fields]; "thrombosis" [All Fields] AND "prenatal diagnosis" [All Fields]; "dural sinus malformation" [All Fields] AND "antenatal diagnosis" [All Fields]; "dural sinus thrombosis" [All Fields] AND "prenatal diagnosis" [All Fields]; "prenatal diagnosis" [All Fields] AND "cerebral thrombosis" [All Fields]; "dural sinus" [All Fields] AND "fetal brain" [All Fields]; "dural sinus malformation" [All Fields] AND "fetal" [All Fields]; "dural sinus malformation" [All Fields] AND "torcular herophili"

Results

Case Report

Case 1

A 30-year-old Caucasian woman (gravida 2, para 1) with unremarkable past medical history and uncomplicated pregnancy was referred to our Institution at 36 weeks' GA for an abnormal third trimester US scan (32nd weeks' GA), showing borderline

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Citation: Gentilomo C, Cavaliere E, Manara R, Nosadini M, Sartori S, et al. (2017) Stem-cell Therapy for Parkinson's Disease. Int J Neuro Disord Interv 1: 109. doi: https://doi.org/10.15344/ijndi/2017/109

ventriculomegaly of both lateral and third ventricles with bilateral subcortical atrophy. Foetal MRI (36th weeks' GA) showed a thrombus at a ballooned TH with extension to the superior sagittal sinus and left transverse sinus, bilateral atrophic-degenerative subcortical areas, corpus callosum thinning and fluid subdural collections (Figure 1). A male infant (BW 2900 g, AI 7-9) was delivered by elective caesarean section at 37 weeks' GA. Generalized seizures were observed since the first hours of life. Postnatal brain MRI (3 days of life) confirmed a stable thrombus, the parenchymal ischemic lesions and ventriculomegaly. In the boy, protein C, protein S, antithrombin, and homocystein levels were normal, antiphospholipid antibodies were not detected, and the screen for factor V Leiden and T20210 mutation was negative, while the mother was heterozygous for PTG20210A mutation. He did not receive antithrombotic treatment. Follow-up brain MRI (1 month of age) was unchanged. At 5 years of age the child suffers from severe mental retardation, quadriplegic cerebral palsy and drug-resistant epilepsy.

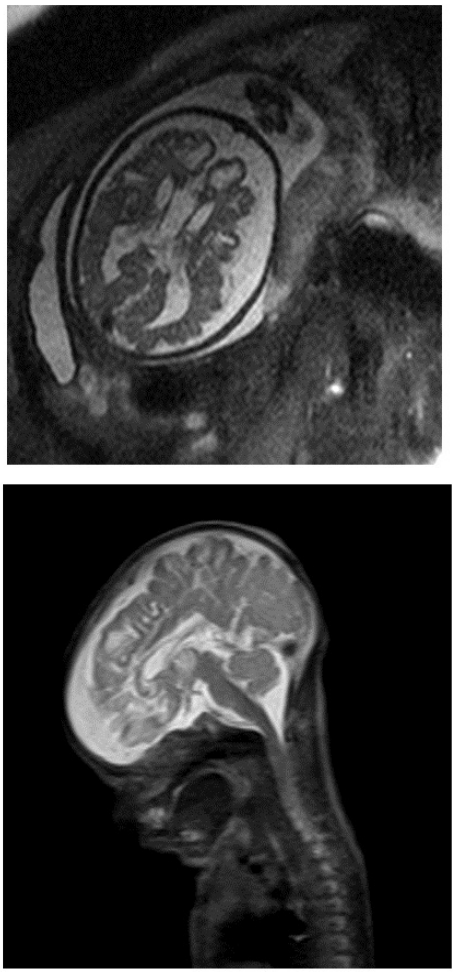

Figure 1: Case 1 – fetal MRI (36 Weeks' GA).
Sagittal and coronal MR T2-W images in a 36 weeks'GA fetus show thrombosis of the lateral transverse sinus and superior sagittal sinus (hyperintense mass) and bilateral subcortical ischemic lesions with corpus callosum thinning.

Case 2

A 37-year-old Caucasian woman (gravida 2, para 0) was referred to our Institution after detection of ventriculomegaly and of a posterior interhemispheric anechoic area at the third trimester US scan (31+5 weeks' GA). A miscarriage one year before was reported. Foetal MRI (32nd weeks' GA), including T1-W, T2-W and DW images, showed dolichocephaly and a massively dilated TH with thrombosis extended to the superior sagittal sinus (Figure 2). Brain development was consistent with gestational age. Follow-up foetal MRI (33+5 weeks' GA) was unchanged. Elective caesarean section at 38+1 weeks' GA delivered a healthy girl (BW 2616 g, AI 9-9). Thrombophilic workup was normal in the newborn and the mother. Transfontanellar US scan at birth showed ventriculomegaly. Postnatal brain MRI (4 days of life) showed partial recanalization of the superior sagittal sinus and TH and disclosed corpus callosum hypoplasia, cortical and subcortical atrophy.

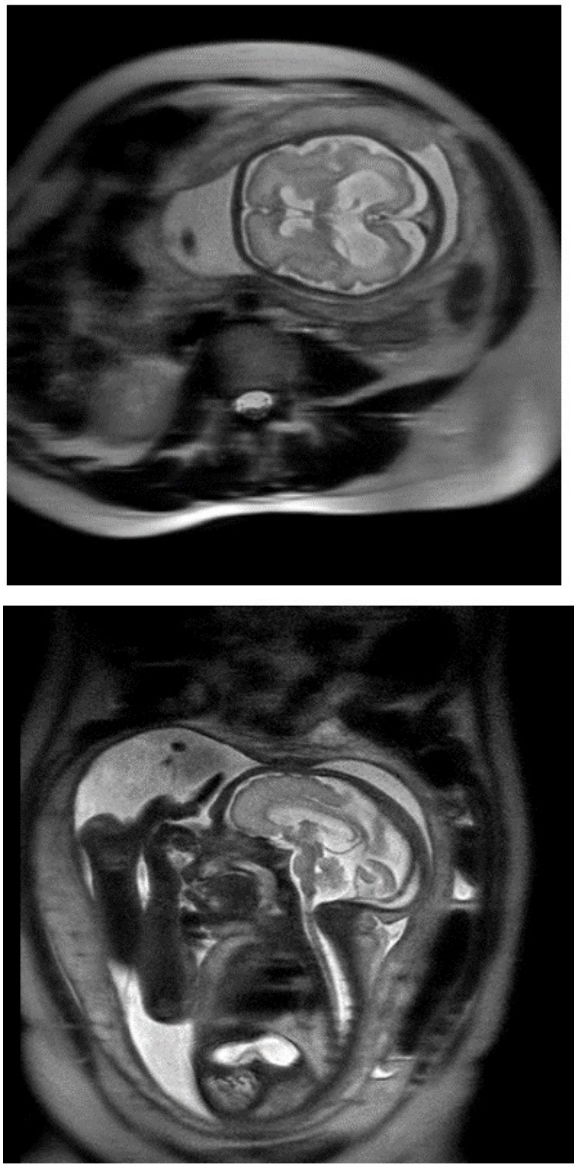

Figure 2: Case 2 – fetal MRI (32 Weeks' GA).
Sagittal and axial MR T2-W images in a 32 weeks'GA fetus show dolichocephaly and a large hyperintense mass inside a markedly ballooned torcular Herophili

Discussion

Sino venous thrombosis is defined by the presence of a thrombus or flow interruption within the cerebral veins or dural sinuses with

or without parenchymal infarction [6]. Antenatal DST is a rare entity that can occur in uncomplicated pregnancies as early as the second trimester. The intracranic abnormality is usually suspected on routine foetal US, but foetal MRI is required for a definitive diagnosis and for better evaluation of possible associated intracranial abnormalities [7,8]. To our knowledge, 66 cases are reported in the literature [2-5, 9-31]. US scan is the usual method of screening, most accessible, for the morphologic evaluation of foetal brain; though, in DST it only shows a nonspecific picture, appearing as a tumour-like median mass in the posterior cranial fossa. By contrast, MRI may allow the differential diagnosis between thrombosis and other intracranial pathologies such as tumors, infections, traumas, and malformations. In the majority of reported cases of foetal DST, brain MRI demonstrated a peculiar picture characterized by a ballooned midline TH with a T2-W iso-hypointense and T1-W hyperintense mass (thrombus) with or without a focal eccentric area of greater T2-W hypointensity (hematoma-haemorrhage); the mass occupied the TH, with variable extension into the superior sagittal sinus [3]. The imaging findings in both our cases were consistent with this description. The cause of foetal tDST is unknown. Early reports described an association with trauma and underlying systemic conditions such as sepsis, meningitis, dehydration, polycythemia, thrombophilia; though, in up to 40% ofthe cases the cause remained unknown [2]. More recently, due to the post-mortem finding of small dural arteriovenous shunts in many cases, an association between foetal tDST and midline DSM with giant pouches (lakes) has been hypothesized3. This type of DSM is thought to arise from the normal ballooning of the sinuses, which occurs during the second trimester (4th-6th month)[1,4,16,27]; it seems to be due to an abnormal development and an uncontrolled growth of the posterior sinuses. It is associated with multiple slow-flow arteriovenous shunting within their walls, which might possibly proceed to larger arteriovenous shunt or to partial or total thrombosis. The thrombosed malformation can spontaneously regress, possibly leading to a good outcome; conversely, in presence of occlusion of all the venous outlets, thrombosis persist and can lead to bilateral venous infarctions or hemorrhages both being risk factors for severe outcome.

It has been suggested that thrombosis of abnormally enlarged TH and posterior dural sinuses is likely secondary to an underlying DSM [3,5].

In both our cases, thrombosis was detected later during the third trimester. Previous US scans were not available, therefore it is impossible to define the time of onset. However, based on the antenatal imaging we can hypothesize that thrombosis could be secondary to a DSM which was detected because of lack of resolution of the cloth [5,4]. We found maternal thrombophilia in one of our patients, but no other associated diseases or triggering conditions, in agreement with most reported cases and previous reviews [14]. Research on prenatal DST reports a wide spectrum of possible outcomes. Of the total 66 patients reported in the literature, 13 were therapeutically aborted. Out of the remaining 53, there were 2 foetal demise and 6 post-natal deaths; among the 45 surviving patients, 6 had neurological sequelae (cerebral palsy, psychomotor delay), while 39 had a favorable outcome with normal development. Based on the above data, the overall mortality rate is 15% (8/53) while a favorable outcome is seen in 87% of surviving patients (39/45). We agree with Laurichesse and colleagues who suggest a spontaneous favorable evolution with decreasing thrombus size, in the absence of parenchymal lesions (infarction or ventricular hemorrhage) and of shunt leading to cardiac congestive failure [14]. Indeed, decrease in thrombus size and recanalization were documented in 92% (35/38) of cases with a favorable outcome (prenatally in 23 foetuses, postnatally in 3, following an initial increase of thrombus size in 4); 4 patients had stable thrombosis; information was not reported in 4; ischemic brain lesions were reported in 3 patients, not known in two (3/36 - 8 %). Among patients with an unfavorable outcome, a follow-up foetal MRI was performed in 5 (3 postnatal deaths, 2 neurological sequelae): in all 3 patients who died, MRI failed to show a decrease in thrombus size, and it variably disclosed other intracranial abnormalities in all but one (ventriculomegaly, intracranial hemorrage, brain compression, dural arterio-venous shunt), but no ischemic lesions. All but two patients who survived with developmental delay had massive ischemic lesions or cerebral atrophy, or progressive ventriculomegaly, despite a decreasing thrombus size. Prognosis was worse for males (30% males in the favorable outcome compared to 75% the unfavorable outcome group). We could not identify other possible prognostic factors in the reviewed literature. Our cases were diagnosed at the third trimester (36 and 32 weeks' GA) and both had a poor neurologic outcome .

Massive ischemic parenchymal lesions were present at diagnosis in one patient and were found postnatally in the second one. A follow- up foetal MRI showed a stable thrombus in patient 2 (33+5 weeks' GA).

In conclusion, our data and literature review suggest that a careful screening of the posterior cerebral fossa at the second trimester routine US is important for early detection of tDST. In case of suspicious images, a prompt referral for foetal MRI supports the diagnosis of DST and allows parental counselling. Results of the present paper suggest that ischemic brain lesions secondary to thrombosis may predict a poor neurologic outcome, while dural arteriovenous shunt or signs of foetal decompensation are associated with an increased risk of foetal demise or post-natal death. Brain lesions may be detected also later during pregnancy, as in our second patient. Follow-up foetal MRI should be continued until the end of pregnancy to allow a better delivery plan and post-natal management.

Competing Interests

The authors declare that they have no competing interests.

Ethical Approval

Our Institutional Review Board approved the retrospective analysis of the patients.

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