



NEONATAL CEREBRAL SINOVENOUS THROMBOSIS AND PROTHROMBOTIC GENETIC MARKERS: ASSOCIATION OR CAUSE?

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BACKGROUND AND AIMS

The association between prothrombotic genetic markers and neonatal cerebral sinovenous thrombosis (CSVT) has been reported in the literature. However, the specific role of these markers is not yet fully understood.

CASE REPORT

PREGNANCY

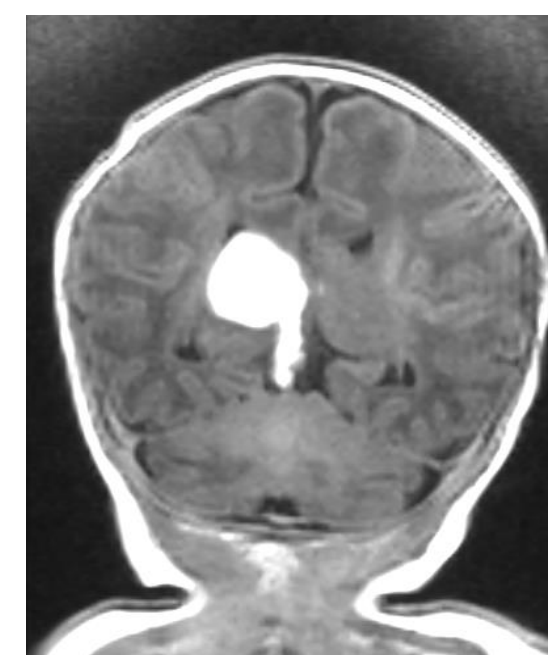
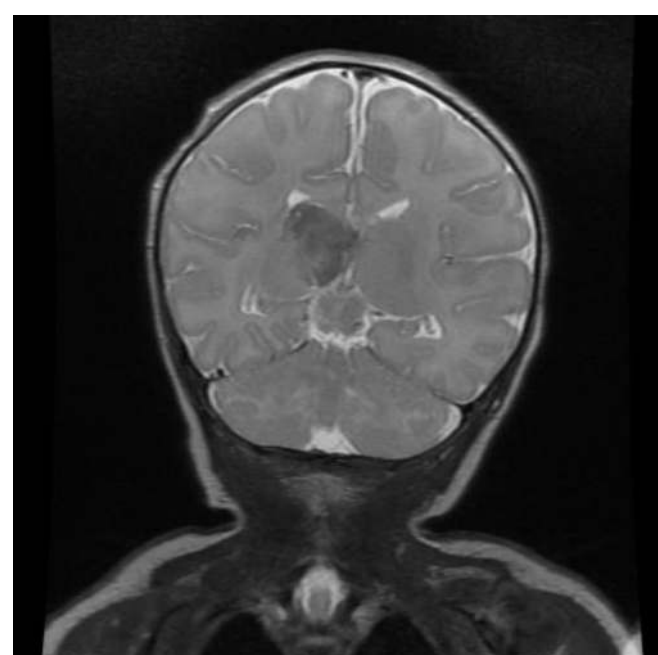
- 30 year-old mother carrier of the **MTHFR 1298A>C** and **PAI-1 5G/4G** allelic variants, **without history of thromboembolism**
- Uneventful pregnancy, 39 weeks and 5/7 days, ultrasounds reported as normal

DELIVERY

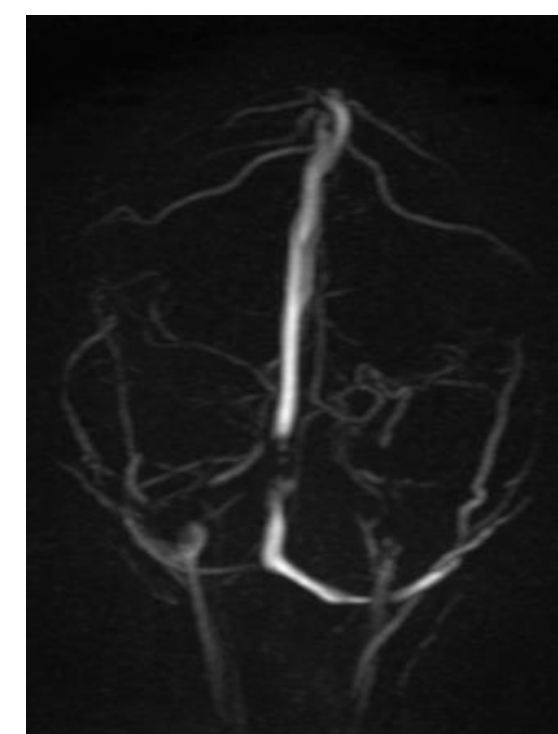
- Male newborn, vaginal birth, **vaccum and forceps extraction**
- Apgar scores **4/8/8**, somatometry in the 10th-50th centile

EVOLUTION

- Mild hypoxic-ischemic encephalopathy (HIE)**
- Good evolution until day 6 of life → **seizures** → phenobarbital
- aEEG monitoring → no subsequent events
- Cerebral ultrasound → **right basal ganglia hemorrhagic infarct**
- Cerebral MRI and magnetic resonance angiography (MRA) → **deep venous thrombosis** (internal cerebral veins, vein of Galen, straight sinus and right lateral sinus) and **thalamic hemorrhage**
- Genetic study of the newborn → **same allelic variants** of the mother
- The clinical course was favorable, with **no paroxysmal events** without anticonvulsant therapy
- Discharged in day 16 of life
- At 2 months → adequate psychomotor development, head circumference in the 50th centile and a normal neurological exam



Figures 1 and 2. Coronal T1 and T2 views showing acute thalamic hemorrhage



Figures 3 and 4. Sagittal T2 showing acute thalamic haemorrhage and MRA showing venous thrombosis

CONCLUSIONS

In our case, there are many factors with possible association to CSVT: HIE, instrumented delivery and prothrombotic allelic variants. In face of unexpected neurological decline, the clinical suspicion was crucial, supported initially by cerebral ultrasound and confirmed by MRI and MRA (gold standard for the diagnosis of CSVT).