A case of congenital hypofibrinogenemia presenting as fetal spontaneous intracranial hemorrhage

Erol O, Ozel KM, Gurses C, Derbent UA
Antalya Training and Research Hospital, Antalya, Turkey

Objective
Fibrinogen is a hexameric glycoprotein essential for coagulation, consisting of two Aα, two Bβ, and two γ chains. Congenital hypofibrinogenemia is a rare bleeding disorder characterized by abnormally low levels of fibrinogen in plasma, generally due to heterozygous mutations in one of the three fibrinogen genes (FGA, FGB, and FGG, coding for Aα, Bβ, and γ chain, respectively). We describe a case of congenital hypofibrinogenemia revealed by the dramatic occurrence of antenatal intracranial hemorrhage, which resulted in severe hydrocephalus.

Methods
Case report.

Results
We present the case of a 36-year-old woman, gravida 3, para1, admitted to antenatal clinic at 32 weeks of gestation. Her medical history was unremarkable. Ultrasonographic examination revealed large anechoic intracranial extra-axial collection causing compression of neuroparenchyma and dilatation of the lateral ventricles (Fig. A and B). Detailed anatomical ultrasound showed no other abnormalities. A detailed evaluation with a fetal MRI could not performed due to maternal obesity (BMI: 54.2 kg/m2). Follow-up scans at 34 and 36 weeks demonstrated adequate fetal growth, and sonographic feature remained unchanged. At 38.3 weeks of gestation, 3140 g male infant was delivered by elective cesarean section. Baby was intubated and ventilated at birth due to poor respiratory efforts. On clinical examination, he had a large head with wide fontanel. Otherwise clinical examination was unremarkable. Plain CT taken postnatally showed extraaxial collection on posterior cerebral fossa with a mass effect (Fig. C and D). The brain architecture was distorted with dilated ventricles. A laboratory examination revealed the following: hemoglobin: 14.3 g/dl, platelet count: 45 000/ml, prolongation of prothrombin time (22.8 s; normal: 11-16s), and partial thromboplastin time (41.2 s; normal 25-33s). The fibrinogen level done by Clauss method was 0.27 g/l (normal: 2-4.5 g/l). Liver and kidney function tests were normal. Peripheral blood film did not reveal any evidence of microangiopathy, hemolysis or infection. In view of the prolonged PT and APTT along with a reduce fibrinogen level, the diagnosis of congenital hypofibrinogenemia was made. Plasma fibrinogen levels of the family members were normal. The epidural hematoma and antenatal detected subdural hematoma were drained by craniotomy. During the first 25 postoperative days, infant treated with haemocomplettan, packed cell transfusion and fresh frozen plasma. The infant died four weeks later after the operation due to necrotising enterocolitis and sepsis.

Conclusion
Congenital fibrinogen deficiency results in a diverse range of bleeding phenotypes. This case is an important addition to the literature as hypofibrinogenemia -related intracerebral hemorrhage is a rare condition.