A case of autosomal dominant inherited Diamond-Blackfan Anemia

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Objective
Diamond-Blackfan Anemia is a rare congenital hypoplastic anemia, characterized by deficiency of the red blood cells, with incidence of seven cases per million of population. Other clinical features are pallor, weakness, congenital malformations like cardiac and skeletal defects and growth retardation. Usually, the condition is discovered in the first year of life. DBA is inherited as an autosomal dominant trait with variable penetrance. The prognosis is generally, good, with regular transfusions and long-term corticosteroid therapy. The objective of this presentation is to mention the importance of vigilant prenatal assessment in pregnancies where there is obstetric history of affected babies with Diamond Blackfan Anemia.

Methods
We present this case report in the light of current literature.

Results
Clinical Presentation: A 29-year-old woman, G3P2, with 2 other children, one healthy boy 5 years old and one 9-year-old girl with Diamond-Blackfan Anemia and infantile epileptic encephalopathy. In her third spontaneous pregnancy, with her consanguineous partner, a CVS was performed at 11 weeks and 5 days, in other department, and the results were normal, showing just a heterozygous state for the infantile epileptic encephalopathy. No chromosomal abnormalities were detected. The sample was not analyzed for the Diamond-Blackfan anemia in view of normal parental phenotype. The first affected baby was considered to have a de novo mutation. During the pregnancy, the mother had several normal scans up to 36 weeks, in our hospital. The mother delivered at 40 weeks and 1 day, a female baby. An emergency cesarean section was performed due to fetal bradycardia following reduced fetal movements. A 3740g baby was born not crying and pale in a very poor condition with signs of bradycardia and hydrops. Baby’s hemoglobin levels were very low (33g/dl) and she required multiple blood products. After 5 days, with cardiovascular and respiratory support in the intensive care, the baby died. Post-mortem examination revealed congenital bone marrow failure with reduction of all three cell lines, especially the erythroid lineage which is suggestive of Diamond-Blackfan anemia. No defects or signs of bacterial or viral infection were detected. The cause of death was multiple organ failure secondary to perinatal asphyxia and severely hypoplastic bone marrow.

Conclusion
The severity of Diamond-Blackfan anemia may vary, even within the same family. Increasingly, individuals with "non-classical" Diamond-Blackfan anemia have been identified. This form of the disorder typically has less severe symptoms. For example, some affected individuals have mild anemia beginning later in childhood or in adulthood, while others have some of the physical features but no bone marrow problems. Therefore, in cases of family history of Diamond-Blackfan anemia, future pregnancies should be screened for this rare genetic condition, even if the parental phenotype is normal.