We report a case of a 31 years-old woman G2P1 refered to our high-risk center because her husband was known for Diamond-Blackfan anemia. He was followed and treated for this condition until adolescence and has been in remission since then. His own father was also affected. Molecular analysis was negative in 2015. 

The patient’s prenatal screening was normal. No prenatal diagnosis was offered because of the absence of a known mutation. Based on the hematologist recommendations, the patient started weekly middle cerebral artery (MCA) Doppler at 16 weeks. At 33 2/7 weeks, the foetus was found to have MCA peak systolic of 83 cm/s (1.78 MoM). Signs of hydrops were not reported. We performed an intrauterine transfusion the next day. The foetus had a pre-transfusion hematocrit of 27% (Hb 89 g/L) and 48ml of blood was transfused. All the subsequent evaluation were normal and the patient delivered a baby girl of 2600g, apgar 2-9-9 by Cesarean section because of breech presentation at 36 4/7 weeks of pregnancy (cord Hb 112 g/L).

Evaluation of the newborn revealed a heterozygote mutation of the RPS19 gene consistent with a diagnostic of Diamond-Blackfan anemia. This mutation was also confirmed for the father.

Diamond-Blackfan anemia is a disorder that primarily affects the bone marrow causing it to malfunction and fail to produce enough red blood cells. This autosomal dominant pathology with variable penetrance affects both genders equally. Over half of all Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial, thumb and urogenital anomalies. Few cases of in utero Diamond-Blackfan anemia have been reported.