Use of High-Performance Liquid Chromatography for Prenatal Diagnosis of α-Thalassemia (Hemoglobin Bart’s) Hydrops Fetalis in Fetal Cord Blood Sample

Jayalakshmi Balakrishna, MD,¹ Men-Jean Lee, MD,² Vijay Shah, MD,² ¹St Luke’s Roosevelt Hospital Centre–Beth Israel Medical Center, ²Beth Israel Medical Center

The gold standard for prenatal diagnosis of α-thalassemia is mutational analysis of fetal cells by chorionic villous sampling or amniocentesis. However, these are time-consuming tests and thus, in the mid-trimester the diagnosis can be made expeditiously by percutaneous umbilical cord blood sampling with high-performance liquid chromatography (HPLC) analysis of the fetal hemoglobins. We present a case of Bart’s hydrops fetalis detected at 21 weeks’ gestation using HPLC from fetal blood obtained by cordocentesis. A 38-year-old Chinese woman presented at 21 weeks’ gestation for a maternal-fetal medicine consultation and prenatal ultrasound evaluation. Both parents were found to have mild microcytic hypochromic anemia and blood was sent for α-thalassemia mutational analysis. Both were found to be α-thalassemia carriers, type 1, which renders 25% of their offspring to be at risk for Bart’s hydrops fetalis. During the initial ultrasound evaluation, there was thickened placenta measuring 9 cm, enlarged fetal heart, and a small pleural effusion. A complete blood count performed on the cord blood sample showed red blood cell count of 0.3 million and hemoglobin of 1.0 g/dL. The cord blood smear showed numerous nucleated red cells. HPLC (Primus Ultra2) showed HgbBart’s 55%, HgbA 39%, and absent HgbF. The presence of 39% of HgbA in the sample from a 21-week fetus was consistent with maternal blood contamination from the placenta. The high level of HgbBart’s with absence of HgbF points towards the absence of α chains. Thus, all HgbA was of maternal origin and not of fetal origin. In conclusion, HPLC can be utilized in immediate prenatal diagnosis of α-thalassemia hydrops fetalis if the clinical situation necessitates it, as in our scenario, since the patient presented at 21 weeks’ gestation, and the legal limits of termination of pregnancy is 24 weeks in the state.