



Recurrent pregnancy loss due to hydrops fetalis caused by alpha thalassemia

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Objective

Hydrops fetalis is defined as fluid collection in at least two body cavities in the fetus. This serious condition occurs due to immune or nonimmune causes. As a result of effective Rh immunization prophylaxis, nonimmune hydrops constitutes approximately 90% of all cases, most commonly due to cardiovascular diseases, chromosomal aberrations and hematological problems. Congenital infections, metabolic, genetic and neurologic disorders, abdominal or thoracic lesions, various congenital anomalies, complication of monochorionic twinning and placental abnormalities are the other causes. Here, we report a patient having repeated pregnancies with nonimmune hydrops fetalis caused by alpha thalassemia.

Methods

A 36-year-old woman, gravida 4, para 3, was referred at 12 weeks of gestation because of increased nuchal translucency. The parents were first-degree cousins. In the patient's history the previous pregnancy had ended in utero with fetal death at 24 weeks of gestation due to hydrops fetalis. Hydrops was first detected at 16 weeks of gestation and amniocentesis had been performed for fetal karyotyping, revealing a normal karyotype. In the index pregnancy, nuchal translucency was measured as 2.5 mm. We recommended chorionic villus sampling for genetic evaluation but the parents declined invasive test. At 17 weeks, ultrasound revealed fetal ascites, pleural effusion and skin edema (Figure 1,2,3). No other structural abnormalities were found. Indirect coombs and serologic tests for infectious diseases such as parvovirus, syphilis, toxoplasmosis and cytomegalovirus were all negative. The pregnancy ended in utero fetal death at 21st weeks of gestation. Pathologic examination was concordant with hydrops and chromosomal analysis of skin biopsy showed a normal karyotype. The mother was anemic with a hemoglobin level of 9,7 g/dl, hematocrit of 29 %, mean corpuscular volume of 81 and mean corpuscular hemoglobin of 27. The patient was counselled with department of Medical Genetics for the suspicion of thalassemia and the parents' carrier status were investigated. According to alpha- globin gene mutation analysis, both parents were found to have thalassemia carrying status.

Results

A 36-year-old woman, gravida 4, para 3, was referred at 12 weeks of gestation because of increased nuchal translucency. The parents were first-degree cousins. In the patient's history the previous pregnancy had ended in utero fetal death at 24 weeks of gestation due to hydrops fetalis. Hydrops was first detected at 16 weeks of gestation and amniocentesis had been performed for fetal karyotyping, which had revealed a normal karyotype. In the index pregnancy, nuchal translucency was measured as 2.5 mm. We recommended chorionic villus sampling for genetic evaluation but the parents denied invasive test. At 17 weeks, ultrasound revealed fetal ascites, pleural effusion and skin edema (Figure 1,2,3). No other structural abnormalities were found. Indirect coombs and serologic tests for infectious diseases such as parvovirus, syphilis, toxoplasmosis and cytomegalovirus were all negative. The pregnancy ended in utero fetal death at 21st weeks of gestation. Pathologic examination was concordant with hydrops and chromosomal analysis of skin biopsy showed a normal karyotype. The mother was anemic with a hemoglobin level of 9,7 g/dl, hematocrit of 29 %, mean corpuscular volume of 81 and mean corpuscular hemoglobin of 27. The patient was counselled by the department of Medical Genetics for the suspicion of thalassemia and the parents' carrier status were investigated. According to alpha- globin gene mutation analysis, both parents were found to have thalassemia carrying status.

Conclusion

Alpha-thalassemia is a hereditary disease that is usually inherited in an autosomal recessive pattern. Therefore, the parents who are alpha-thalassemia carriers have 25% chance of having of a child with major alpha-thalassemia. Among the hemoglobinopathies, alpha-thalassemia is the most common cause of nonimmune hydrops fetalis. Since the incidence of alpha-thalassemia is reported high both in Turkey (XX) and in Balkan region, genetic counseling for alpha-

thalassemia should be suggested to patients having fetuses with nonimmune hydrops fetalis. If both parents are carriers, then genetic counseling together with preimplantation genetic investigation should be considered for future uneventful pregnancies.

