



β-Thalassemia: Early prenatal diagnosis

β-Talasemi: Doğum öncesi erken tanı

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ABSTRACT

A case of beta-thalassemia major diagnosed by fetal DNA analysis on a chorion biopsy during the 11th week of gestation is described. A 30 year-old Gravida 2, Parite 1 patient whose firstborn died of beta thalassemia major (Cd8/A) when 2 years old, was admitted to our clinic on her 10th week of pregnancy. Chorionic villus sampling was performed and resulted as beta thalassemia major (Cd8/Cd8).

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ÖZET

Bu vakada 11 hafta gebelik sırasında koryon villus biyopsisiyle beta talasemi majör tespit ettik. Hasta 30 yaşında ve bu ikinci gebeliğiydi. İlk bebeği 2 yaşında beta talasemi major nedeniyle ölmüş. Biz bu vakayı 10 hafta gebelikte değerlendirdik. Beta talasemi major tespit ettik ve gebeliği sonlandırdık.

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A case

A case of beta-thalassemia major diagnosed by fetal DNA analysis on a chorion biopsy during the 11th week of gestation is described.

Thalassemia is one of the major monogenic disorders affecting a large section of the world's population. In β-thalassemia major, the genetic defects are mostly due to point mutations in both alleles of the beta-globin genes. The affected babies are healthy up to age 3-6 months. Then patients gradually develop severe anemia, and regular blood transfusions are necessary to maintain normal growth and

development. Treatment is very expensive and only palliative. Therefore, prevention of birth of the thalassemic children through prenatal diagnosis is the major way to control the spread of the disease (Li et al., 2006).

If the molecular defect in the disease gene is known, direct DNA analysis using a polymerase chain reaction (PCR) or restriction fragment length polymorphisms (RFLP) probe is possible. Examples of analysis currently diagnosed by direct detection include α- and β-thalassemia, sickle cell anemia and cystic fibrosis (Koçak et al., 1998).

A 30 year-old Gravida 2, Parite 1 patient whose firstborn

died of beta thalassemia major (Cd8/A) when 2 years old, admitted to our clinic on her 10th week of pregnancy. Chorionic villus sampling was performed and resulted as beta thalassemia major (Cd8/Cd8). Carrier detection of thalassemia in asymptomatic patients can be achieved by different methods such as red blood cell indices mean cell volume (MCV), haemoglobin electrophoresis, polymerase chain reaction (PCR) technique and erythrocyte osmotic fragility test (EOFT). PCR was used for DNA analysis. Prenatal diagnosis of β -thalassemia can be done by mutation analysis of the DNA isolated from chorionic villus sampling, provided the mutations have already been determined. The prenatal diagnosis revealed affected foetuses with homozygous status of beta-

thalassemia major (both alleles of mutant variety Cd8/Cd8). The case mutation analysis showed that both parents were carriers of b-globin gene mutation Mother had β -thalassemia mutation (Cd8/A) and father had β -thalassemia mutation (Cd8/A). Fetus was terminated with misoprostol.

Thalassemia is one of the most common health problems in word, where the incidence of abnormal globin gene is rather high. This contributes to the high incidence of infants with a severe form of thalassemia homozygous. These infants will grow and live with a poor quality of life. To improve the efficacy of prevention of the new cases, all pregnant women in the high prevalence areas should be tested for the carrier status (Wanapiraka et al., 1998).

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