SHORT COMMUNICATION

BETA-TThALASSEMIA AND CHROMOSOMAL ABERRATIONS

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Introduction

Beta-thalassemia is the most common hereditary disease in Iran and more than 2 million carriers of the β-thalassemia mutant gene are living in this country.1,2 About 110 mutant genes have been recognized all over the world1,2 of which 21 have been identified in the Iranian population. The mutant genes and their frequencies vary greatly in different parts of Iran.1,2 However, it seems that at least five more undetected mutant genes exist.

Materials and Methods

In a 13-year period, 2500 individuals were analyzed for β-thalassemia genes and 472 prenatal tests were performed in our center.

Results

Our data show that 130 fetuses had inherited both parental mutant genes (thalassemia major)3, 220 fetuses carried one mutant gene (thalassemia minor), and 95 inherited neither mutant gene (normal); 3 fetuses carried hemoglobin D, and one carried hemoglobin S. The fetal condition regarding one allele (50% prenatal diagnosis) was only determined in 9 cases and 14 cases remained inconclusive to both alleles.

We also performed a cytogenetic study in 46 of these cases (25 chorionic villus samples and 21 amniotic fluid cultures), of which three cases revealed chromosomal aberrations.

Discussion

In the current studies, since both unbalanced karyotypes belong to fetuses of mothers with advanced age. Considering that 2 (12.5%) of the fetuses of 16 mothers who were karyotyped because of advanced age, showed unbalanced chromosomal aberrations, we would like to...
Figure 1. Heterochromia of irises and the white forelock, which is fairly noticeable in the picture, are characteristic for Waardenburg syndrome.

emphasize the necessity of performing cytogenetic studies for fetuses of all parous women with advanced age, including those being tested for β-thalassemia.

References