

## SHORT COMMUNICATION

# BETA-THALASSEMIA AND CHROMOSOMAL ABERRATIONS

Mohammad-Hassan Karimi-Nejad MD<sup>1</sup>, Hossein Najmabadi PhD, Maryam Zangeneh MSc, Roxana Karimi-Nejad MSc

*Karimi-Nejad Pathology and Genetic Center, Tehran, Iran*

### Introduction

**B**eta-thalassemia is the most common hereditary disease in Iran and more than 2 million carriers of the  $\beta$ -thalassemia mutant gene are living in this country.<sup>1,2</sup> About 110 mutant genes have been recognized all over the world<sup>1,2</sup> of which 21 have been identified in the Iranian population. The mutant genes and their frequencies vary greatly in different parts of Iran.<sup>1-2</sup> 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  $\beta$ -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)<sup>3</sup>, 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.

The first case was a male fetus of a  $\beta$ -thalassemia mother married to her consanguineous cousin. She was 32-year-old, gravida 6, para 2, abortion 3. Chorionic villus sample revealed 46, XY,t (4,14) (q28;q31), which was also confirmed on cultured amniotic cells. Cytogenetic study of the parents showed that the translocated chromosome was inherited from the mother.

The second case was a male fetus of a 37-year-old woman gravida 4 para 3. The chorionic villus sample of her fourth pregnancy was analyzed and karyotyped for  $\beta$ -thalassemia and revealed 47, XY, +21 (Down's syndrome).

The third case, a 39-year-old woman gravida 6 para 5 with 3 dead and 2 living children, was the paternal cousin of her 44-year-old husband. Her first three pregnancies resulted in 3 boys, who expired shortly after birth. The outcome of the fourth pregnancy was a boy with  $\beta$ -thalassemia major. We performed prenatal test for their fifth fetus in July 1991, which revealed that the fetus was a carrier of the  $\beta$ -thalassemia minor gene. Postnatal examination confirmed the prenatal test, but he also had Waardenburg syndrome, inherited from the mother (Figure 1).

Molecular and cytogenetic analyses of amniotic fluid culture of the sixth pregnancy showed a  $\beta$ -thalassemia minor female fetus with trisomy of chromosome 18.

### 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

•Correspondence: M.H. Karimi-Nejad MD, Karimi-Nejad Pathology and Genetic Center, Tehran, Iran. P.O.Box: 14665/154, Fax: +9821-807 7487, Email: [mhkariminejad@mavara.com](mailto:mhkariminejad@mavara.com).



**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  $\beta$ -thalassemia.

### References

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