TABLE Association of specific β thalassaemia mutations and chromosomal haplotypes in the Turkish population.

<table>
<thead>
<tr>
<th>Haplotype</th>
<th>IVS-1, nt 110</th>
<th>Uncharacterised</th>
</tr>
</thead>
<tbody>
<tr>
<td>HincII</td>
<td>HindIII</td>
<td>HincII</td>
</tr>
<tr>
<td>x</td>
<td>Gy</td>
<td>AY</td>
</tr>
<tr>
<td>I</td>
<td>+</td>
<td>−</td>
</tr>
<tr>
<td>III</td>
<td>+</td>
<td>−</td>
</tr>
<tr>
<td>VI</td>
<td>−</td>
<td>+</td>
</tr>
<tr>
<td>VII</td>
<td>+</td>
<td>−</td>
</tr>
<tr>
<td>X</td>
<td>−</td>
<td>+</td>
</tr>
</tbody>
</table>

*Oligonucleotide analysis excluded the presence of the β*39 and the IVS-1, nt 6 mutations.

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References


A new case of (Y;1) balanced reciprocal translocation in an infertile man with Hodgkin’s disease

The proband was a 31 year old man with no children after two years of marriage. His two brothers, uncles, and aunts were fertile. There was no history of infectious disease or trauma. He was 1.73 m tall, weighed 65 kg, and had no dysmorphic features. Physical examination showed slight testicular hypotrophy (12 and 15 ml) and a varicocele. The

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male genitalia and secondary sex characteristics were normal. FSH plasma levels were raised (19-4 IU/l), but LH (11 IU/l) and prolactin (7.0 µg/l) were normal and plasma testosterone level was decreased. No information on sperm count was obtained.

Biopsies from the left and right testicles showed similar features: a decrease in the diameter of the seminiferous tubules with dystrophic lesions. Numerous spermatogonia and pachytene spermatocytes only were seen but no secondary spermatocytes.

Chromosome investigations were carried out on peripheral blood cultures. PHA stimulated lymphocytes were examined after R and C banding. All 37 R banded metaphases examined showed a reciprocal translocation t(Y;1)(q21;p13) (fig 1a, b). C banding confirmed that the breakpoint in the Y chromosome had occurred within the heterochromatic segment (fig 1c). The proband is being treated for Hodgkin's disease and refused further investigations. His relations also refused to have a blood sample taken.

Y:acrocentric translocations appear not to affect the phenotype and they may be transmitted as a chromosome variant through both males and females. There was no increased incidence of infertility or spontaneous abortions in the families of the patients described by Smith et al and they may be associated with a normal Y. These translocations apparently involve only heterochromatic material consisting of highly repeated DNA sequences which are not transcribed.

Y:non-acrocentric translocation is commonly associated with primary infertility and, in those cases where family studies were carried out, the abnormality had arisen de novo. The mechanism leading to infertility in these translocations is not known, but abnormal segregation at meiosis may be involved in maturation arrest. The translocated Y induces an abnormal sex vesicle and two opposing results are possible. Either the Y segment not included in the sex vesicle is not normally inactivated, or the autosome translocated onto the Y is partly inactivated by a spreading effect.2

Our case is the second report of Y;1 translocation. The first one described a four month old male infant with t(Y;1)(q11;q21) who showed severe psychomotor retardation, infantile spasms, and hypotonia. These clinical abnormalities, like the Hodgkin's disease in our case, may be coincidental to the chromosomal rearrangement.

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Distal trisomy 14q

At least 20 cases of trisomy for the distal part of 14q have been reported.1 3 The main clinical signs of this trisomy are common to all types of autosomal imbalance. They include prenatal growth retardation, physical and psychomotor retardation, wide sutures and fontanelles, a prominent forehead, hypertelorism, a protruding upper lip, a high arched palate, abnormal ears, micrognathia, heart defects, and cryptorchidism. Because of this, it is difficult to diagnose distal trisomy 14q clinically. The phenotypic heterogeneity might be caused by both the genetic composition of the long arm of chromosome 14 and genetic heterogeneity in the cases described. The present report describes a case of trisomy 14q24–q32 resulting from a paternal insertion (4;14).

The proband was the product of a term first pregnancy of unrelated healthy 25 year old parents. Her birth weight was 2370 g and length 45 cm. The following clinical features were noted at birth: haematoma of the eyelids, dystopia opisthotonus, spasticity and hyperreflexia of the lower extremities.

Severe physical and psychomotor retardation was evident at nine months. Her weight was 5000 g, length 61 cm, and