Unbalanced whole-arm translocation der(1;13) in hematologic malignancies

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Abstract

Whole-arm chromosome translocations involving the long arm of chromosome 1 are nonrandom aberrations in hematologic malignancies that commonly involve acrocentric chromosomes. Among them, unbalanced whole-arm translocations between chromosomes 1 and 13 are relatively rare cytogenetic aberrations and has been reported in both lymphoid and myeloid neoplasms.

Keywords
chromosome 1; chromosome 13; translocation; hematologic malignancies

Clinics and pathology

Disease
Myeloproliferative disorders, multiple myeloma and lymphoid malignancies

Phenotype/cell stem origin
Reported in diverse hematologic disorders

Etiology
Different factors like constitutional fragility of the 1q heterochromatin, cytotoxic drugs, ionizing radiation and/or oncogenic viruses are suspected to be implicated in the origin of 1q rearrangements.

Epidemiology
Reported at least in 20 cases (Table 1); median age 52 years (range 20-86), balanced sex ratio (11M/9F). Found in: chronic myeloproliferative disorders (8 of the 20 available cases): polycythemic myelofibrosis (PPMF) in 1 case, essential thrombocytopenia (ET) in 1 case, chronic myeloid leukemia (CML) in 1 case; myelodysplastic syndromes (MDS) in transformation in 3 patients as well as in acute myeloid leukaemia (2 patients), multiple myeloma (4 patients) and in lymphoid malignancies (8 patients).

Evolution
Whether karyotypic changes associated with extra copies of 1q are primary events or they are induced during disease evolution as a side effect of cytotoxic treatments is unclear.

May be found as a sole anomaly in chronic myeloproliferative disorders (Andrieux et al, 2003; Tanaka et al, 2006), indicating that der(1;13) might be a primary change in myeloid disorders.

Occurred as part of complex karyotypes in multiple myeloma and lymphoproliferative malignancies, suggesting that 1q abnormalities may be secondary events in these diseases representing clonal evolution associated with natural disease evolution.

Prognosis
It is likely that the prognosis depends on the patient diagnosis in myeloid malignancies (chronic disease versus acute leukemia). Prognosis in multiple myeloma and lymphoid malignancies is uncertain.
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G-banded partial karyogram of bone marrow cells showing the der(1;13)(q10;q10) chromosome. Fluorescence in situ hybridization with Vysis (Abbott Molecular) LSI 1p36 (red)/1q25 (green) and LSI Rb (red) probes showing an extra green signal located on 1q25 on the der(1;13)(q10;q10) chromosome (arrow)

Cytogenetics

**Cytogenetics morphological**

Presents as -13, + der(1;13)(q10;q10); less frequently as der(1;13)(p10;q10) or der(1;13)(q10;p10)

**Genes involved and proteins**

Genes involved are unknown; the region 1q21-1q32 has been suggested to contain oncogenes that are involved in disease pathogenesis

Result of the chromosomal anomaly

**Fusion protein**

**Oncogenesis**

Acquired whole-arm chromosome translocations with involvement of the 1q heterochromatin are accompanied by genomic imbalances in hematologic malignancies. The chromosome 1 pericentromeric heterochromatin is a notoriously an unstable chromosomal region that is involved in diverse chromosomal rearrangements leading to gene dosage abnormalities.
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<table>
<thead>
<tr>
<th>Chromosome anomalies</th>
<th>Disease</th>
<th>Sex/Age</th>
<th>Ref.</th>
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<tbody>
<tr>
<td><strong>MYELOPROLIFERATIVE DISORDERS</strong></td>
<td></td>
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</tr>
<tr>
<td>1. 46,XY,der(1;13)(q10;p10);t(11;19)(p13;q13)</td>
<td>PPMF</td>
<td>M; 64</td>
<td>8</td>
</tr>
<tr>
<td>2. 50,XX,der(1;13)(q10;13),+8t(8;16)(p22;q12)</td>
<td>AML</td>
<td>M; 60</td>
<td>16</td>
</tr>
<tr>
<td>3. 48,XY,del(13)(q10),t(5;10),p10</td>
<td>MDS progressed to AML</td>
<td>M; 17</td>
<td>1</td>
</tr>
<tr>
<td>4. 46,XY,der(1;13)(q10);del(17)(q12)</td>
<td>MDS in transformation</td>
<td>M; 49</td>
<td>3</td>
</tr>
<tr>
<td>5. 47,XX,der(1;13)(q10;10)</td>
<td>MDS in transformation</td>
<td>F; 58</td>
<td>7</td>
</tr>
<tr>
<td>6. 46,XX,del(3)(q22);q15</td>
<td>FA, developed AML</td>
<td>M; 49</td>
<td>1</td>
</tr>
<tr>
<td>7. 46,XY;+t(22q;q11)</td>
<td>CML in transformation</td>
<td>F; 20</td>
<td>18</td>
</tr>
<tr>
<td>8. 46,XX,del(13)(q10),t(13;17)(q10;q11)</td>
<td>at diagnosis</td>
<td>ET, developed granulocytic sarcoma</td>
<td>F; 75</td>
</tr>
</tbody>
</table>

| MULTIPLE MYELOMA |
|-------------------|---------|------|
| 1. 53,XX,del(13)(q10) | MM | F; 54 | 14 |
| 2. 47,35,XY;+t(12;15)(p11;q15) | MM | M; 44 | 5 |
| 3. 38,XX,del(13)(q10) | MM | F; 7 | 6 |
| 4. 44,XX,del(13)(q10),t(13;17)(q10;q11) | MM | M; 44 | 19 |

| LYMPHOID-NEOPLASMS |
|---------------------|---------|------|
| 1. 46,Xadd(Xq28),der(1;13)(q10;10)(q11;10) | Mature B-cell neoplasm | F; 35 | 12 |
| 2. 44,45,XY,del(17)(q25),+t(12;13)(q25;q13) | DLBCL | M; 49 | 2 |
| 3. 80,XXY,del(13)(q10;11p) | Mature B-cell neoplasm, NOS | M; 86 | 4 |
| 4. 47,XXY,del(13)(q10),del(13)(q10),del(18)(q11),del(20)(q11),add(5) | Mature T- and NK-cell neoplasm | M; 43 | 10 |
| 5. 47,XXY,del(13)(q10),del(13)(q10),del(18)(q11),del(20)(q11),add(5) | Follicular lymphoma | M; 30 | 1 |
| 6. 47,XXY,del(13)(q10),del(13)(q10),del(18)(q11),del(20)(q11),add(5) | DLBCL | M; 43 | 10 |

The acquisition of the long arm of chromosome 1 results in trisomy of the whole-arm of chromosome 1 and partial monosomy of the involved chromosome. Duplication of the chromosome segment of 1q11-1q32 is commonly observed in these rearrangements, indicating that certain chromosome 1 regions, especially 1q21-1q32 might harbor pathogenetically relevant oncogenes. The unbalanced nature of the der(1;13)(q10;q10) indicates that the gain of 1q may play an important role in neoplastic transformation and/or disease progression. Although a der(1;13)(q10;q10) translocation has been reported in various neoplastic conditions, such as multiple myeloma and lymphoma, this translocation is also observed in both chronic and acute myeloid disorders. The observation of this anomaly was closely associated with leukemic transformation in myeloid malignancies suggesting that der(1;13)(q10;q10) might be a rare but nonrandom primary change in these disorders preceding or accompanying disease evolution.

References


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