Leukaemia Section
Short Communication

der(1;14)(p10 or q10;p10 or q10)
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Abstract
Review on der(1;14)(p10 or q10;p10 or q10) translocation, with data on clinics

Clinics and pathology

Disease
Myelodysplastic syndrome (MDS), myeloproliferative neoplasm (MPN), acute myelogenous leukemia (AML), acute lymphoblastic leukemia (ALL), multiple myeloma (MM) and lymphoma.

Phenotype/cell stem origin
Suggested involvement of a pluripotent stem cell.

Epidemiology
Most of patients (9/14) had a diagnosis of myeloid disorder: AML (2 patients), MDS (2 cases) and chronic myeloproliferative disorders (5 cases). A few other cases included ALL (2 patients), MM (1 case) and 2 cases with lymphoid malignancies. The male sex is prevalent (9:5); the median age at diagnosis is 59 years (range 10-74 years) (Table 1).

Prognosis
The prognostic effect of an extra 1q chromosome in myeloid disorders may be variable (indolent clinical course in MPD vs a distinct possibility of transformation and poor prognosis in MDS). The prognosis of patients with other diseases is unknown (sporadic cases reported).

Cytogenetics

Cytogenetics morphological
Whole arm chromosome translocation showing 2 normal copies of chromosome 1, only 1 normal chromosome 14 and a der(1;14) resulting in complete 1q trisomy. The centromeric breakpoints were confirmed only in sporadic cases, revealing an unique centromere derived from chromosome 14 (Busson-Le Coniat et al.,1999; Djordjevic et al., 2005; Fogu et al., 2013). The preservation of both chromosome 1 and 14 centromeres was not confirmed.

Additional anomalies
Sole anomaly in half cases; found in association with del(5q), del(7q), del(12p), del(20q), t(9;22)(q34;q11) in myeloid malignancies and with t(8;14)(q24;q32) in Burkitt lymphoma.

Result of the chromosomal anomaly

Fusion protein
Oncogenesis
The unbalanced 1q whole-arm translocation with the recipient acrocentric 14 chromosome results in pure trisomy of the long arm of chromosome 1. The main consequence of 1q trisomy is a genomic imbalance that may account for a proliferation advantage of the neoplastic clone through a gene dosage effect.
The consequent deregulation of several genes results in alteration of the balance between proliferation and cell death, suggesting that gene deregulation is the main mechanism of oncogenesis in 1q rearrangements, similar to numerical aberrations, such as trisomy 8 observed in various hematologic malignancies.

Table 1. Reported cases with der(1;14) centromere translocations involving 1p10 or 1q10.

<table>
<thead>
<tr>
<th>Sex/Age</th>
<th>Karyotype</th>
<th>Disease</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>M/59</td>
<td>46.XY,t(1;14)(q10;q10)</td>
<td>Post-PV MDS</td>
<td>Swolin et al., 1986</td>
</tr>
<tr>
<td>F/28</td>
<td>46.XX,t(1;14)(q10;q10),der(19)t(1;19)(q23;p13)</td>
<td>ALL</td>
<td>Nylund et al., 1994</td>
</tr>
<tr>
<td>M/54</td>
<td>46.XY,der(1;14)(q10;p10)</td>
<td>NHL</td>
<td>Hashimoto et al., 1995</td>
</tr>
<tr>
<td>F/64</td>
<td>46.XX,der(1;14)(q10;p10),del(1)(q22)</td>
<td>Post-PV MF</td>
<td>Andrieux 2003</td>
</tr>
<tr>
<td>F/59</td>
<td>47.X,add(X)(p22),+7,t(8;14)(q24;q22)/47,der(12)(q10)</td>
<td>DL</td>
<td>Chan et al., 2003</td>
</tr>
<tr>
<td>F/11</td>
<td>46.XX,t(1;14)(q10;q10),del(9)(q34),add(17)(p11)</td>
<td>RAEB</td>
<td>Imashuku et al., 2003</td>
</tr>
<tr>
<td>M/69</td>
<td>46.XY,der(1;14)(p10;q10),del(12)(p12)</td>
<td>RAEBt</td>
<td>Harada et al., 2004</td>
</tr>
<tr>
<td>F/73</td>
<td>46.XX,der(1;14)(q10;q10)</td>
<td>PV</td>
<td>Zamora et al., 2004</td>
</tr>
<tr>
<td>M/65</td>
<td>46.XY,t(1;14)(q10;q10),del(5)(q13q33),-8,+9,del(11)(q14),del(20)(q11),der(21)(p11)</td>
<td>RARS</td>
<td>Barouk-Simonet et al., 2005</td>
</tr>
<tr>
<td>M/73</td>
<td>46.XY,t(1;14)(q10;q10)</td>
<td>ALL</td>
<td>Adeyinka et al., 2007</td>
</tr>
<tr>
<td>M/74</td>
<td>46.XY,t(1;14)(q10;q10)</td>
<td>MM</td>
<td>Adeyinka et al., 2007</td>
</tr>
<tr>
<td>M/43</td>
<td>46.XY,t(1;14)(q10;q10),del(14)(q22)</td>
<td>MPD</td>
<td>Adeyinka et al., 2007</td>
</tr>
<tr>
<td>M/43</td>
<td>46.XY,t(1;14)(q10;q10),del(7)(q22)</td>
<td>CMoL</td>
<td>Djordjevic et al., 2008</td>
</tr>
<tr>
<td>M/43</td>
<td>46.XY,t(1;14)(q10;q10),del(7)(q22)</td>
<td>AML- M4</td>
<td>Lee et al., 2008</td>
</tr>
</tbody>
</table>

Unbalanced 1q whole-arm translocation resulting in der(14)t(1;14)(q11-12;p11) in myelodysplastic syndrome Cytogenet Genome Res 2012;136(4):256-63

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