t(1;19)(p13;p13.1)

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Abstract

Review on t(1;19)(p13;p13.1), with data on clinics.

Clinics and pathology

Disease
Myeloid malignancies.

Epidemiology
Only 5 cases to date.

Clinics
2 patients with acute myeloid leukemia (AML): a 1 year-old infant with M5a AML (Tchinda et al; 2002) and a 36 year-old male patient with AML-M1 (Ma et al; 2000), one 77-year-old female patient with post-polycythemic myelofibrosis (Suh et al; 2009) and 2 myelodysplastic syndrome (MDS) patients with refractory anemia with ringed sideroblasts (RARS): a 21 year-old female with a suspicion of Fanconi anemia 11 years before diagnosis of RARS (Tchinda et al; 2002) and a 60 year-old female patient (Suh et al; 2009).

Prognosis
Limited data; death occurred 8 months after diagnosis in the case with AML of infant (Tchinda et al; 2002); prognosis may be variable (chronic vs acute disease).

<table>
<thead>
<tr>
<th>Sex</th>
<th>Age</th>
<th>Chromosomal anomalies</th>
<th>Diagnosis</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>M</td>
<td>40-46,XY,+der(1)t(1;19)(p13;p13.1)</td>
<td>AML-M1</td>
<td>Ma et al; 2000</td>
</tr>
<tr>
<td>2</td>
<td>F</td>
<td>47,XX,+der(1)t(1;19)(p13;p13.1),der(10)inv(10)(p25q25),t(10;11)(q25;q25),der(11)t(10;11)</td>
<td>AML-M5a</td>
<td>Tchinda et al; 2002</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>47,XX,+der(1)t(1;19)(p13;p13.1)</td>
<td>MDS</td>
<td>Tchinda et al; 2002</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>47,XX,+der(1)t(1;19)(p13;p13.1)</td>
<td>MDS</td>
<td>Suh et al; 2009</td>
</tr>
<tr>
<td>5</td>
<td>F</td>
<td>47,XX,+der(1)t(1;19)(p13;p13.1)</td>
<td>Post PV-MF</td>
<td>Suh et al; 2009</td>
</tr>
</tbody>
</table>

Table 1. Reported patients with der(1)t(1;19)(p13;p13.1).

Abbreviations: F, female; M, male; AML, Acute myeloid leukemia; MDS, myelodysplastic syndrome; post PV-MF, post polycythemic myelofibrosis
Cytogenetics

Cytogenetics morphological

The translocation presents as + der(1)t(1;19)(p13;p13) in all the 5 known cases.

Additional anomalies

Sole anomaly in 4 of the 5 cases; complex anomalies in an infant patient with AML-M5a.

Genes involved and proteins

Note

The unbalanced rearrangement described as der(1)t(1;19)(p13;p13.1) has rarely been reported, may be found in sporadic cases of patients with myeloid neoplasms such as AML, MDS and polycythemia vera (PV). The possible role of such unbalanced translocation in disease pathogenesis needs to be determined and the genes implicated in this rearrangement still remain unknown. While no gene rearrangements were detected in this translocation, it is possible that ELL, the gene located on chromosome 19p13.1, may be involved in at least some cases.

ELL is the MLL partner gene in t(11;19)(q23;p13.1) translocation resulting in MLL-ELL fusion that is found exclusively in patients with myeloid malignancies. Similar to t(11;19)(q23;p13.1), the unbalanced der(1)t(1;19)(p13;p13.1) may constitute a specific entity in myeloid neoplasms, occurring mainly in adults.

References


This article should be referenced as such: