Leukaemia Section

Short Communication

i(Xq10) in female patients

Tatiana Gindina

R.M. Gorbacheva Research Institute of Pediatric Oncology Hematology and Transplantation at First Saint-Petersburg State Medical University named I.P.Pavlov, Saint-Petersburg, Russia / tatgindina@gmail.com

Published in Atlas Database: February 2017
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/iXq10FemaleID1493.html
Printable original version: http://documents.irevues.inist.fr/bitstream/handle/2042/68752/02-2017-iXq10FemaleID1493.pdf
DOI: 10.4267/2042/68752

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2018 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Abstract

ABSTRACT
Review on i(X)(q10) in female patients.

KEYWORDS
Chromosome X; Acute lymphoblastic leukemia; Diffuse large B-cell lymphoma; Burkitt lymphoma; Chronic myelogenous leukemia; Myelodysplastic syndrome; Acute myeloid leukemia.

Clinics and pathology

Disease

i(X)(q10) occurs in a variety of hematologic malignancies, in both myeloid and lymphoid disorders, including ALL, NHL, AML, MDS and CML.

Chronic myeloid leukemia (CML): 2 patients (Lyall and Garson, 1978; Karrman et al., 2007).

Myelodysplastic syndrome (MDS) was diagnosed in 2 patients (Wilkens et al, 1998; Kearns et al., 2004).

Acute myeloid leukemia (AML): there were 8 patients with AML-M2, six of them had translocation t(8;21)(q22;q22) (Shiraishi et al, 1982; Minamihisamatsu et al, 1988; Tien et al, 1988; Kwong et al, 1993; Ma et al, 1997; Paskulin et al, 1998).

Acute lymphoblastic leukemia (ALL): was diagnosed in 6 patients. Three of them with relapse of ALL after chemotherapy (CT) (Arthur et al., 1982; Gindina T, own cases, table 1,#18, #19), two patients had unbalanced translocation t(1;19)(q23;p13) (Pui et al, 1990; Gindina T, table 1,
i(Xq10) in female patients

#18, one patient had translocation t(4;11)(q21;q23) (Arthur et al, 1982).

**Follicular lymphoma** (FL): 3 patients (Horsman et al, 2001; Fan and Rizkalla 2003).

**Burkitt lymphoma** (BL) was diagnosed in 2 patients (Goyens et al., 1993; Trcic et al., 2010).

**Diffuse large B-cell lymphoma** (DLBCL): 2 patients (Bloomfield et al., 1983; Dave et al, 2002).

**Mature B-cell neoplasm**: 2 patients (Ueda et al, 1997; Adeyinka et al, 2007).

**Epidemiology**

I(X)(q10) has been observed in female patients with a median age of 27 years (range, 0.9 to 54 years).

### Table 1. Reported cases with i(X)(q10)

<table>
<thead>
<tr>
<th>Pt</th>
<th>Age, Sex</th>
<th>Disease</th>
<th>Karyotype</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>40, F</td>
<td>CML</td>
<td>46, X, i(X)(q10), del(1)(q22), t(17;17)(q12; q25)</td>
<td>Karrman et al, 2007</td>
</tr>
<tr>
<td>2</td>
<td>?, F</td>
<td>CML</td>
<td>46, X, i(X)(q10), t(9;22)/47,idem,+der(22)t(9;22)</td>
<td>Lyall &amp; Garson, 1978</td>
</tr>
<tr>
<td>3</td>
<td>?, F</td>
<td>MDS</td>
<td>47, X, i(X)(q10), +21</td>
<td>Kearns et al, 2004</td>
</tr>
<tr>
<td>4</td>
<td>?, F</td>
<td>MDS</td>
<td>47, XX, +i(X)(q10)</td>
<td>Wilkens et al, 1998</td>
</tr>
<tr>
<td>5</td>
<td>54, F</td>
<td>AML (M2)</td>
<td>45, X, -X, del(9)(q22)/6/46, X, i(X)(q10)/6</td>
<td>Han et al, 2002</td>
</tr>
<tr>
<td>6</td>
<td>?, F</td>
<td>AML (M2)</td>
<td>46, X, i(X)(q10), t(8;21)(q22; q22)</td>
<td>Kwong et al, 1993</td>
</tr>
<tr>
<td>7</td>
<td>33, F</td>
<td>AML (M2)</td>
<td>46, X, i(X)(q10), t(8;21)(q22; q22)/5/46,XX/3</td>
<td>Ma et al, 1997</td>
</tr>
<tr>
<td>8</td>
<td>?, F</td>
<td>AML (M2)</td>
<td>47, X, i(X)(q10), +8, t(8;21)(q22; q22)</td>
<td>Minamihisamatsu et al, 1988</td>
</tr>
<tr>
<td>9</td>
<td>45, F</td>
<td>AML (M2)</td>
<td>88, XXY, +i(X)(q10), x2; (1)(q10), dup(2)(q11q21)x2, del(3)(q21), del(5)(q13q35)x2, -6, -6, -9, -12, (12)(q10), -15, -15, -17, -17, -21, add(21)(q22), add(22)(q12)x2, +mar</td>
<td>Pang et al, 2015</td>
</tr>
<tr>
<td>10</td>
<td>30, F</td>
<td>AML (M2)</td>
<td>46, X, i(X)(q10), t(8;21)(q22; q22)/46, idem, del(9)(q13q22)</td>
<td>Tien et al, 1988</td>
</tr>
<tr>
<td>11</td>
<td>?, F</td>
<td>AML (M2)</td>
<td>46, XX, t(8;21)(q22; q22)/46, idem, i(X)(q10)[7]/45, idem, -X[5]</td>
<td>Paskulin et al, 1998</td>
</tr>
<tr>
<td>12</td>
<td>40, F</td>
<td>AML, (M2)</td>
<td>46, X, i(X)(q10), t(8;21)(q22; q22)</td>
<td>Shiraishi et al, 1982</td>
</tr>
<tr>
<td>13</td>
<td>10, F</td>
<td>AML, NOS</td>
<td>46, X, i(X)(q10)</td>
<td>Debiec-Rychter et al, 1985</td>
</tr>
<tr>
<td>14</td>
<td>?, F</td>
<td>ALL</td>
<td>47, X, i(X)(q10), t(10;11)(p13; q13), +19</td>
<td>Heerema et al, 1998</td>
</tr>
<tr>
<td>15</td>
<td>0.9, F</td>
<td>ALL</td>
<td>44, X, i(X)(q10), -7, -8, der(10)t(8;10)(q11;p13), der(19)t(1;19)(q23;p13)</td>
<td>Pui et al, 1990</td>
</tr>
</tbody>
</table>
As a single anomaly, isochromosome Xq was described only in two cases of myeloid malignancies (Debiek-Rychter et al, 1985; Wilkens et al, 1998). Double extra i(X)(q10) was present in 5 patients (Ueda et al, 1997; Wilkens et al, 1998; Horvman et al, 2001; Dave et al, 2002; Pang et al, 2015). Additional chromosome abnormalities were observed in 26/28 patients. i(X)(q10) has been demonstrated in six cases with RUNX1 / RUNX1T1 AML (Shiraishi et al, 1982; Minamihisamatsu et al, 1988; Tien et al, 1988; Kwong et al, 1993; Ma et al, 1997; Paskulin et al, 1998), two cases with TCF3 / PBX1 ALL (Pui et al, 1990; Gindina T, table 1, #18) and one case with KMT2A / AFF1 (Arthur et al, 1982). Associated in combination with other isochromosomes in 4 patients (Dave et al, 2002; Adeyinka et al, 2007; Trcic et al, 2010; Pang et al, 2015), del(9q) in 3 patients (Tien et al, 1988; Han et al, 2002; Gindina T, table 1, #19) and extra chromosome 21 in 2 (Kearns et al., 2004; Pui et al., 1992). In lymphomas, i(X)(q10) is usually part of a complex karyotype.

**Result of the chromosomal anomaly**

**Fusion protein**

**Oncogenesis**
As a result of the formation of the isochromosome, there is the loss of a normal X chromosome, and the structural abnormality leads to monosomy for Xp and trisomy for Xq. It is not known whether the overexpression of a proto-oncogene (or other gene) or the deletion of a tumor-suppressor gene from the isochromosome contributes to development or proliferation of tumor cells in these cases.

References


Ma SK, Au WY, Kwong YL, Lam CK, Liang RH, Chan LC. Hematological features and treatment outcome in acute myeloid leukemia with i(16;21). Hematol Oncol. 1997 May;15(2):93-103


This article should be referenced as such: