Leukaemia Section
Short Communication

der(18)t(1;18)(q10-25;q11-23)

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Abstract
Review on der(18)t(1;18)(q10-25;q11-23) translocation, with data on clinics and cytogenetics.

Clinics and pathology

Disease
Acute myeloid leukemia (AML), acute lymphocytic leukemia (ALL), multiple myeloma (MM) and lymphoid neoplasms

Phenotype/cell stem origin
Pluripotent.

Etiology
Myeloid neoplasms: a 64 year old female with refractory anemia and 4 patients with AML (1 female infant with Down syndrome and 3 adults (2 males, 1 female; aged 21 to 66 years). Acute lymphoblastic leukemia: a 2 years old female infant with Down syndrome and 3 adults (2 males and 1 female; aged 43 to 70 years). Multiple myeloma: female prevalence (1 male, 10 females) and 12 patients with various lymphoid malignancies: (6 males, 6 females; aged 34 to 70 years)

Epidemiology
32 cases to date; 11 male and 21 female patients aged 1-70 yrs (Table 1)

Partial karyotypes with der(18)t(1;18)(q10;q11)
<table>
<thead>
<tr>
<th>Sex/Age</th>
<th>Chromosome anomalies</th>
<th>Disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>M</td>
<td>46,XX,+8,der(18)(q11.2;q21.3)</td>
<td>AML-M5</td>
</tr>
<tr>
<td>M</td>
<td>46,XY,del(9)(p22);add(12)(q11.2)</td>
<td>T-cell</td>
</tr>
<tr>
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<td>46,XY,der(18)(q12.1;q21.3)</td>
<td>ALL</td>
</tr>
<tr>
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<td>46,XY,del(18q);+19</td>
<td>B-cell</td>
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<tr>
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<td>MM</td>
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<td>MM</td>
</tr>
<tr>
<td>16</td>
<td>46,XX,del(9)(p22);add(12)(q11.2)</td>
<td>MM</td>
</tr>
</tbody>
</table>

Table 1: Part 2 - Abbreviations:

| 17 | F | 53.XX,+3,+5,+7;inv(8)(p21q24),del(79)(p22),+11,del([14](q24),der(18)(1;18)(q12;q23),+19,+20.unc | MM |
| 22 | M | 58 | 47.XY,+3,+5,+8,der(14)(10;14)(q33;q32),der(18)(1;18)(q18;q23) | FL |
| 23 | M | 66 | 45-47,X,-X,t(2;4)(p23;q33),add(6)(p25),+12,der(18)(1;18)(q18;q23) | AML |
| 24 | F | 57 | 46.XX,der(X)(X),q28,q12-21,46.XX,der(18)(1;18)(q18;q23) | FL |
| 25 | F | 39 | 45.XX,del(1)(p21q25),t(8;14)(q24;q32),der(18)(1;18)(q18;q23) | BL |
| 26 | F | 44 | 46.XXX,der(18)(1;18)(q23),45-46.XX,der(X)(1;X)(q12;q28) | FL |
| 27 | M | 49 | 46.dup(X)(p22),Yt(8;14)(q24;q32),der(18)(1;18)(q12;q21),47,idem,add(15)(q26),+16 | PT LPD |
| 28 | F | 50 | 50.XX,+X,+5,+7;inv(8)(p21q24),del(79)(p22),+12,del(13)(q14),t(14;18)(q32;q21),der(18)(1;18)(q22;q23) | DLBCL |
| 29 | M | 48 | 48.XY,+X,+7,del(10)(q22q24),der(18)(1;18)(q11;q23),48,idem,(t;8;14)(q42;q32),del(11)(p13p15),dup(12)(q15q21) | FL |
| 30 | M | 46 | 46.XY,del(1)(q24),der(6)t(6;8)(q15;q13),der(6)(6;15)(p22;q11),+del(7) | MCL |
| 31 | M | 34 | 46.XXY,der(2)t(2;14)(p12q32),+7,del(1)(q13q24),der(14)(10;14)(q12 or p11,p11)(t;8;14)(q24;q23),-21,der(22)(t;21;22) | BCL |
| 32 | F | 44 | 80-82.XXX,add(1)(p13),add(2)(q37),add(3)(p25),inv(3)(p21p25),-4,del(3)(q11q33),del(7)(q31q35),-8,-12,-13,-14,-15,-16,del(17)(p12),der(18)(1;18) | ATLL (HTLV-l+)

**Prognosis**
Unknown; likely unfavorable in cases with complex karyotypes.

**Cytogenetics**

**Cytogenetics morphological**

Found as -18, + der(18)(t;1;18)(q10-25;q11-23) with 2 normal chromosomes 1, a normal chromosome 18 and a der(18) chromosome. Breakpoints in 1q were clustering to 1q21-23; the 18q breaks occurred mostly in 18q21-23 region.

**Additional anomalies**

May be associated with known anomalies such as t(8;21)(q22;q22) or +8 in AML and t(9;22)(q34;q11) in ALL; part of complex karyotypes in MM. Associated with 14q32 rearrangements, (8 cases); +7 (4 cases), -X+X (5 cases) and chromosome X anomalies (3 cases) in lymphoid malignancies.

**To be noted**

The unbalanced der(18)(t;1;18)(q10-25;q11-23) results in partial trisomy for the 1q segment and loss of genes from 18q leading to gene dosage abnormalities. May be detected in both hematologic neoplasms and lymphoid malignancies. Found in association with known primary anomalies in acute leukemias, indicating that this aberration is mostly a secondary event representing clonal evolution. Frequent chromosomal change in multiple myeloma and lymphoid neoplasms, where it is part of complex karyotypes associated with tumor progression advanced disease.

**References**


Katzenberger T, Ott G, Klein T, Kalla J, Müller-Hermelink HK, Ott MM. Cytogenetic alterations affecting BCL6 are predominantly found in follicular lymphomas grade III with a diffuse large B-cell component. Am J Pathol. 2004 Aug;165(2):481-90


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