**CASE REPORTS in HAEMATOLOGY**  
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**Translocation t(8;12)(q13;p13) in a case with acute leukemia of ambiguous lineage**

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### Clinic

- **Age and sex**: 2 year(s) old female patient.
- **Previous History**: no preleukemia
  - no previous malignant disease
  - no inborn condition of note
- **Organomegaly**: hepatomegaly (3cm from CRB); splenomegaly (3cm from CRB); enlarged lymph nodes (Mediastinal lymph nodes); no central nervous system involvement

### Blood

- **WBC**: 254.4 x 10^9/l; Hb: 6.8 g/dl; platelets: 86 x 10^9/l;
- **Bone marrow**: 89% (of infiltration by L2 lymphoblasts)

### Cytopathology

- **Cytology**: Ambiguous lineage acute leukemia
- **Immunophenotype**: Blast cells were positive for T lineage antigens: CD2, CD7, cCD3; B lineage antigens: CD19 and cCD79a and myeloid antigens: CD13 and CD33. A minor (10%) myeloid blast population was also detected among the leukemic cells expressing MPO and CD117. Other positive markers were CD45, CD34, HLA-DR whereas CD10 and TdT were negative.
- **Rearranged Ig Tcr**: -
- **Pathology**: -
- **Electron microscopy**: -
- **Precise diagnosis**: acute leukemia of ambiguous lineage

### Survival

- **Date of diagnosis**: 04-2006
- **Treatment**: 12-ALLIC/ BFM-PROTOCOL
- **Complete remission was obtained**
- **Treatment related death**: -
- **Relapse**: -
- **Status**: Alive 07-2007
- **Survival**: 14 month(s)

### Karyotype

- **Sample**: Bone marrow; culture time: 24 h; banding: G banding
- **Results**: 46,XX,t(8;12)(q13;p13)[16]/46,XX[4].
- **Karyotype at relapse**: -
- **Other molecular cytogenetics techniques**: Fluorescence in situ hybridization (FISH) with painting probes (WCP 8, WCP 12) and LSI ES Dual Color Translocation Probes (TEL/AML1 and AML1/ETO) (Vysis, Inc.)
- **Other molecular cytogenetics results**: ish t(8;12)(q13;p13)(WCP8+,WCP12+,TEL+EETO,WCP8+,WCP12+,ETO+,TEL-). FISH analysis with TEL/AML1 probe revealed that the gene ETV6 is in the derivate 8 and it is not involved in the translocation.
- **Other molecular studies techniques**: RT-PCR
- **results**: BCR-ABLp210, BCR-ABLp190, MLL-AF4, MLL-AF9, MLL-ENL, E2A-PBX1 and ETV6-RUNX1 were negative.
Partial GTG banded karyotype showing t(8;12)(q13;p13).

FISH with painting probes for chromosomes 8 (green) and 12 (red).
FISH with TEL/AML1 and AML1/ETO probes.

Comments
To our knowledge four cases of t(8;12)(q13;p13) have been reported in the literature. Three of them were described in childhood acute leukemia. The fourth case was described during disease progression in acute myelomonocytic leukemia with t(11;19). It was suggested that t(8;12) might play an important role in the relapse and lead to a poor prognosis. Our patient presented a bad response to prednisone and was considered of high risk group. At present she remains in complete remission.

Internal links
Atlas Card  t(8;12)(q13;p13)

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