**Case Report Section**

**Paper co-edited with the European LeukemiaNet**

**der(1;18)(q10;q10) in a pediatric patient with cytopenias**

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Published in Atlas Database: July 2013

Online updated version : http://AtlasGeneticsOncology.org/Reports/der0118q10q10CytZamecID100071.html

DOI: 10.4267/2042/52081

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**Abstract:** Case report on der(1;18)(q10;q10) in a pediatric patient with cytopenias.

### Clinics

**Age and sex**

9 years old male patient.

**Previous history**

No preleukemia, no previous malignancy, no inborn condition of note, no main items

**Organomegaly**

No hepatomegaly, no splenomegaly, no enlarged lymph nodes, no central nervous system involvement.

### Blood

- **WBC:** 4.5 X 10^9/l (neutrophils = 55%, eosinophils = 2%, lymphocytes = 32%, monocytes = 10%, atypical lymphocytes = 1%)
- **HB:** 11.6g/dl
- **Platelets:** 149X 10^9/l
- **Blasts:** 1%

**Bone marrow:** Bone marrow studies showed myeloid maturation arrest, intermittent neutropenia with normal erythropoiesis and megakaryocytes.

### Cyto-Pathology Classification

**Cytology**

NA

**Immunophenotype**

Not done

**Diagnosis**

MDS – unclassified

### Survival

**Date of diagnosis:** 03-2012

**Treatment:** No therapy

**Treatment related death:** no

**Relapse:** no

**Status:** Alive

**Last follow up:** 06-2012

**Survival:** 12 months

### Karyotype

**Sample:** Bone marrow

**Culture time:** 24h

**Banding:** GTG

**Results**

46,XY,+1,der(1;18)(q10;q10) [5]/ 46,XY [25]

**Other molecular cytogenetics technics**

Fluorescence in situ hybridization applying the LSI 1p36/1q25 probe (Abbott).

**Other molecular cytogenetics results**

Two signals for 1p36 locus with 3 signals for 1q25 locus in 20% of bone marrow cells.
der(1;18)(q10;q10) in a pediatric patient with cytopenias

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(A) Partial karyotype of the patient showing the der(18)(1;18)(q10;q10). (B) C-banded partial karyotype showing the der(1;18)(q10;q10) chromosome. (C) Fluorescence in situ hybridization with LSI 1p36/1q25 probe (Abbott) showing 3 copies of the 1q25 locus in two nuclei (green signal; arrow).

Comments

This study reports the presence of an unbalanced translocation between chromosome 1 and chromosome 18 in a pediatric patient with persistent thrombocytopenia and intermittent neutropenia. Unbalanced translocations involving the long arm of chromosome 1 and different partners are recurrent cytogenetic abnormalities, mainly reported in myeloid neoplasms. Centromeric fusion between chromosome 1 and chromosome 18, leading to a gain of 1q and loss of 18p, is rarely observed. This abnormality is relatively restricted to myelodysplastic syndromes and myeloproliferative disorders, indicating that gain of 1q and/or loss of 18p should be relevant for neoplastic transformation in these diseases.

References


Wan TS, Ma SK, Au WY, Chan LC. Derivative (1;18)(q10;q10): a recurrent and novel unbalanced translocation involving 1q in myeloid disorders. Cancer Genet Cytogenet. 2001 Jul 1;128(1):35-8


This article should be referenced as such: