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Case Report Section

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

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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

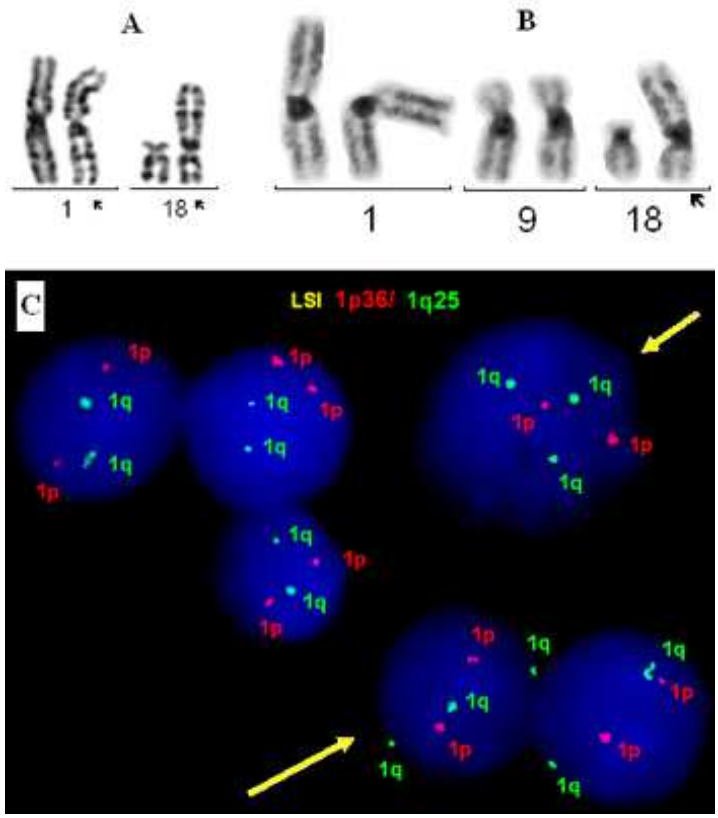
Culture time: 24h

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.



(A) Partial karyotype of the patient showing the der(18)t(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 (Abott) 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 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 myloproliferative disorders, indicating that gain of 1q and/or loss of 18p should be relevant for neoplastic transformation in these diseases.

References

Sawyer JR, Swanson CM, Koller MA, North PE, Ross SW. Centromeric instability of chromosome 1 resulting in multibranched chromosomes, telomeric fusions, and "jumping translocations" of 1q in a human immunodeficiency virus-related non-Hodgkin's lymphoma. Cancer. 1995 Oct 1;76(7):1238-44

Sawyer JR, Swanson CM, Wheeler G, Cunniff C. Chromosome instability in ICF syndrome: formation of micronuclei from multibranched chromosomes 1 demonstrated by fluorescence in situ hybridization. Am J Med Genet. 1995 Mar 27;56(2):203-9

Polito P, Canzonieri V, Cilia AM, Gloghini A, Carbone A, Gaidano G. Centromeric instability of chromosome 1 resulting in multibranched chromosomes, telomeric fusions, and "jumping translocations" of 1q in a human immunodeficiency virus-related non-Hodgkin's lymphoma. Cancer. 1996 Sep 1;78(5):1142-4

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

Caramazza D, Hussein K, Siragusa S, Pardanani A, Knudson RA, Ketterling RP, Tefferi A. Chromosome 1 abnormalities in myeloid malignancies: a literature survey and karyotype-phenotype associations. Eur J Haematol. 2010 Mar;84(3):191-200

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