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Case Report Section

Short Communication

A new case of Acute Myeloid Leukemia with semi-cryptic t(7;21)(p22;q22)

Tatiana Gindina, Ildar Barkhatov, Elmira Boychenko, Irina Garbuzova, Maria Vlasova, Elena Nikolaeva, Irina Petrova, Varvara Ovechkina, Tatiana Shorstova

I.P. Pavlov State Medical University, R.M. Gorbacheva Memorial Institute of Children Hematology and Transplantation, Cytogenetic Laboratory, Saint Petersburg, Russia (TG, IB, MV, EN, IP, VO, TS), City Children's Hospital No.1, Saint Petersburg, Russia (EB, IG)

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Clinics

Age and sex
13 years old male patient.

Previous history
No preleukemia, no previous malignancy, no inborn condition of note.

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

Blood

WBC: 2,5X 10^9/l
HB: 11,3g/dl
Platelets: 278.000X 10^9/l
Blasts: 0%
Bone marrow: 56% (Hypocellular bone marrow, the number of blasts increased up to 56%. Blasts had round shape with a rounded nucleus and looped chromatin structure, with large blue nucleoli. The cytoplasm of blasts was blue and narrow without granularity. Part of the blasts had irregular shape with a bean-shaped nucleus and bright large nucleolus. Their cytoplasm was moderate or abundant, gray-blue with small azurophilic granules. Myelopoiesis: myeloid dysplasia, some cells have giant nuclei, scanty specific granularity, dissociation in the maturation of the nucleus and cytoplasm. Granulocytic population comprised approximately 26% of bone marrow cells. Erythropoiesis was represented by normoblasts. Megakaryocytopoiesis was decreased with hypolobular megakaryocytes).

Cyto-Pathology Classification

Cytology
Acute myelomonocytic leukemia (AML), M4

Immunophenotype
Positive for CD45dim, CD33, CD56, CD34, CD7, CD117, CD13, CD71, HLA-DR; additionally, there is abnormal coexpression of CD4 and CD11b.

Rearranged Ig Tcr
Not performed.

Pathology
Not performed.

Electron microscopy
Not performed.

Diagnosis
AML M4 (FAB), AML with multilineage dysplasia (WHO).

Survival

Date of diagnosis: 12-2011

Treatment: Induction therapy included AIE (cytarabine/idarubicin/etoposide) and HAM (high-dose cytarabine 3 g/m^2/mitoxantrone).

Complete remission was achieved after FLAG regimen (fludarabine, cytarabine, granulocyte-colony-stimulating factor) applied as consolidation therapy.

Complete remission: Complete hematological and cytogenetic remissions were achieved in April 2012.

Treatment related death: no.
Relapse: no.

Status: Alive. Last follow up: 04-2012.

Survival: 5 months.

Karyotype

Sample: Bone marrow aspirate.

Culture time: 24h, without stimulating agents.

Banding: GTG

Results

Analysis of 20 metaphase cells revealed an abnormal male karyotype.
46,XY,add(21)(q22)[19]/46,XY,idem,del(5)(q13q31)[1].

Other molecular cytogenetics technics

Multicolour fluorescence in situ hybridisation (mFISH) using the 24XCyte Human Multicolor FISH Probe kit (MetaSystems, Germany) and FISH with LSI ETV6(TEL)/RUNX1 (AML1) Dual Color Translocation Probe Set (Abbott Molecular, USA) were performed.

Other molecular cytogenetics results

The semi-cryptic translocation t(7;21) was revealed in all metaphases and split of the RUNX1 gene was detected in the interphase nuclei (three red signals).
46,XY,t(7;21)(p22;q22)[20].ish t(7;21)(RUNX1+;RUNX1+)[20].

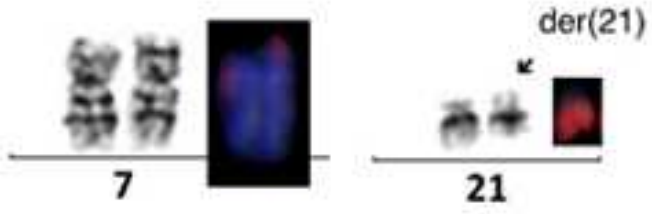
Other Molecular Studies

Technics:

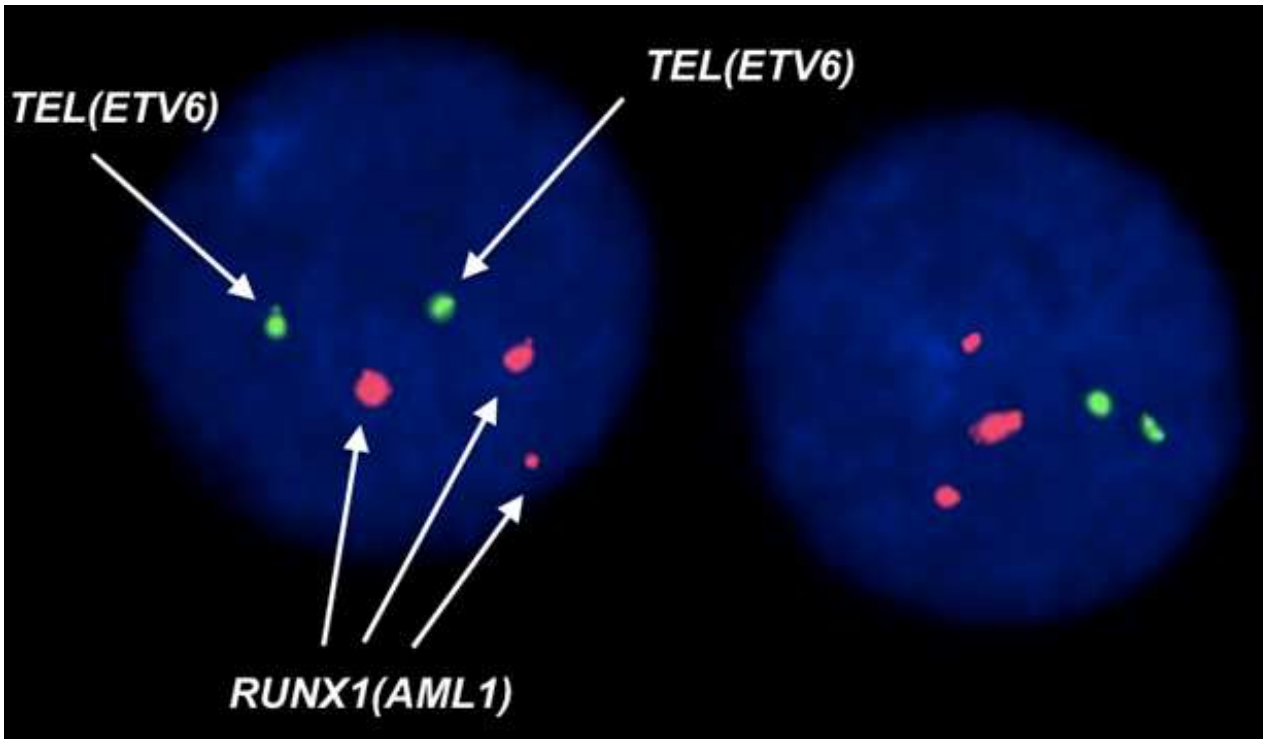
ASO-PCR, RFLP.

Results:

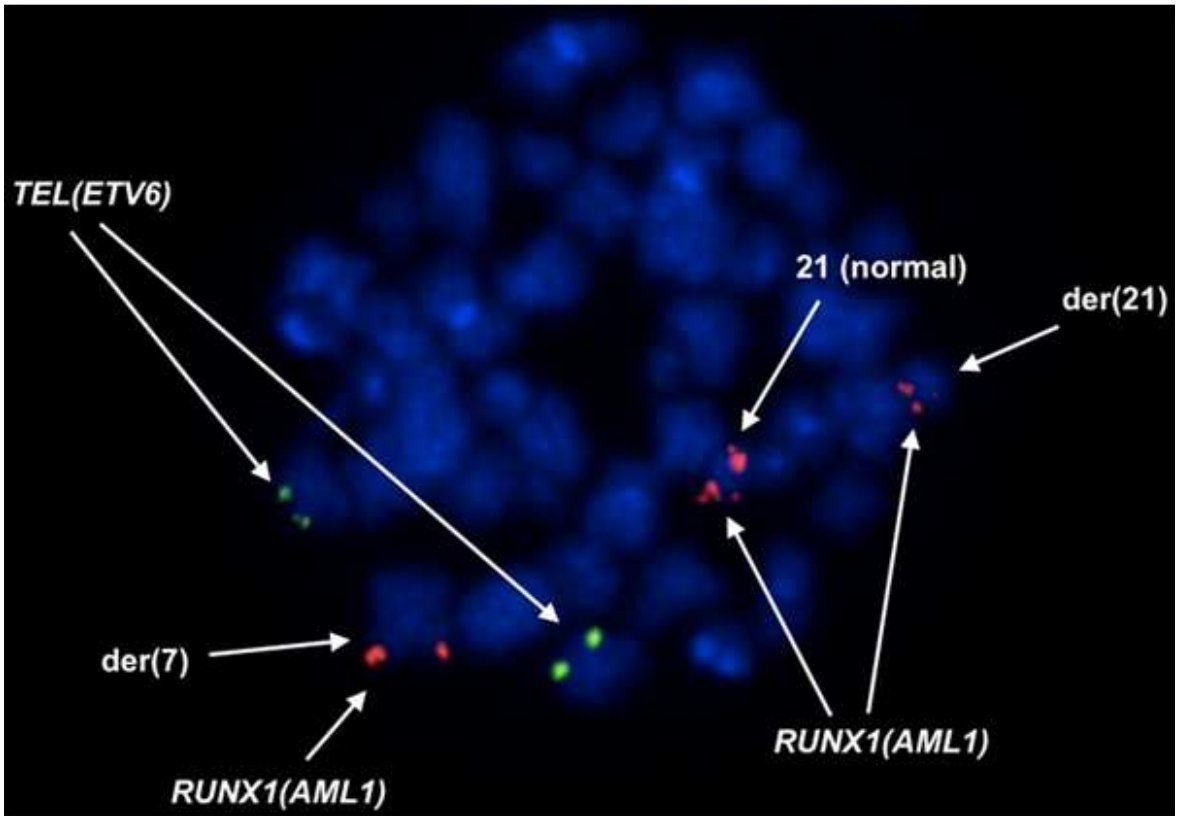
Mutations in NPM1 and FLT3 genes (ITD&D835) were not detected.



G-banded and partial mFISH karyograms showing t(7;21).



Interphase FISH with three signals of RUNX1 (red) and two signals of ETV6 (green) genes.



FISH with LSI ETV6/RUNX1 probe showing the red signals of RUNX1 on der(7) chromosome, der(21) chromosome and on normal chromosome 21. Two normal signals (green) of ETV6 are seen on the chromosomes 12.

Comments

Here, we report a new case of AML M4 with semi-cryptic t(7;21)(p22;q22). As known the translocation t(7;21)(p22;q22) is a rare recurrent abnormality in MDS and AML that results in a RUNX1-USP42 fusion as described previously (Paulsson et al., 2006). Although all four patients with t(7;21) revealed a similar cytogenetic lesion, they varied in their clinicopathological features: of the three adults, the first one presented with RAEB-2, the second AML M5, the third AML M0 and the 7-year-old child had AML M0 too. All adults had chemosensitive disease, whereas the child had refractory AML following initial induction therapy and relapsed following allogeneic bone marrow transplantation. Current case of 13-year-old boy diagnosed with acute myelomonocytic leukemia also had evidence of persistent AML after initial course of treatment; he achieved complete hematological and cytogenetic remissions after FLAG.

Call for Collaborations

MD, PhD Tatiana Gindina
Cytogenetic Laboratory, R.M. Gorbacheva Memorial Institute of Children Hematology and Transplantation, I.P. Pavlov State Medical University, Saint-Petersburg, Russia, Leo Tolstoi Str., 6/8, 197022

Telephone: + 7 (812) 233 12 43
Fax: + 7 (812) 233 96 01

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