Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL

INIST-CNRS

Case Report Section

A new case of adult Acute Myeloid Leukemia with isolated tetrasomy 4p

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Published in Atlas Database: May 2016
Online updated version : http://AtlasGeneticsOncology.org/Reports/Tetra4pRichebourgID100085.html
Printable original version : http://documents.irevues.inist.fr/bitstream/handle/2042/68536/05-2016-Tetra4pRichebourgID100085.pdf
DOI: 10.4267/2042/68536

Abstract

Case report on a new case of adult Acute Myeloid Leukemia with isolated tetrasomy 4p.

Clinics

Age and sex
65 years old male patient.

Previous history
No preleukemia
No previous malignancy
No inborn condition of note
No relevant medical history

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

Blood

WBC: 17,49X 10^9/l
HB: 8,9g/dl
Platelets: 117X 10^9/l
Blasts: 15%
Bone marrow: Not performed

Cyto-Pathology Classification

Phenotype
Acute Myeloid Leukemia

Immunophenotype
f peripheral blood blast cells CD34+,HLA-DR+,CD13+,CD33+,CD117+,CD11c+,CD4+

Rearranged Ig Tcr
Not performed

Pathology
Not performed

Electron microscopy
Not performed

Diagnosis
Acute Myeloid Leukaemia, NOS

Survival

Date of diagnosis
08-2015

Treatment
IDAC induction followed by 4 HDAC consolidations

Treatment related death : no
Relapse : no

Status: Alive

Last follow up: 02-2016

Survival: 6 months +

Karyotype

Sample: peripheral blood

Culture time
24 colchicine over-night, 24 hours and 48 hours with GM-CSF

Banding : GTG

Results
47,XY,+i(4)(p10)[6]/46,XY[14]

Other molecular cytogenetics technics
Fluorescent in situ hybridization using KMT2A Break Apart probe (Cytocell LPH013) and FGFR3-IGH Dual Fusion probe (Cytocell LPH030)

Other molecular cytogenetics results
ish i(4)(p10)(FGFR3++)[3].nuc ish(KMT2Ax2)[99/100]

Other Molecular Studies

Technics
RT-PCR assays on blood sample demonstrating the absence of BCR-ABL1, RUNX1T1-RUNX1, CBFb-MYH11 and PML-RARA transcripts; PCR assays on blood sample demonstrating the presence of NPM1 mutation and the absence of FLT3-ITD mutation.

Comments

We here present a new case of supernumerary i(4)(p10) in a case of male adult AML. According to the literature, this is the seventh case reported in a context of myeloid neoplasm, the last one described in the atlas by Desangles et al.,2013 (Hagemeijer et al., 1981; Hoo et al., 1995; Chen et al., 1999; Soriani et al., 2010). Despite the rarity of this abnormality, it is interesting to note that common features arise from the reviewed cases (table 1). At first, there is a strong association with male sex, since six of the seven patients were male.
Secondly, among the AML cases, it seems there is an association with a monocytic differentiation: indeed three of the four precedent cases were classified into acute myelomonocytic leukemia and in the present case, despite the absence bone marrow evaluation and the absence of peripheral monocytosis, the expression of CD4 and CD11c on blast cells demonstrated by flow cytometry is consistent with this observation. Regarding the prognostic significance of this abnormality, the limited number of cases and the absence of long term follow-up don't allow to draw any formal conclusion. The only case of relapse reported concerns an AML4 case harboring a double supernumerary isochromosome 4p (Soriani et al., 2010).

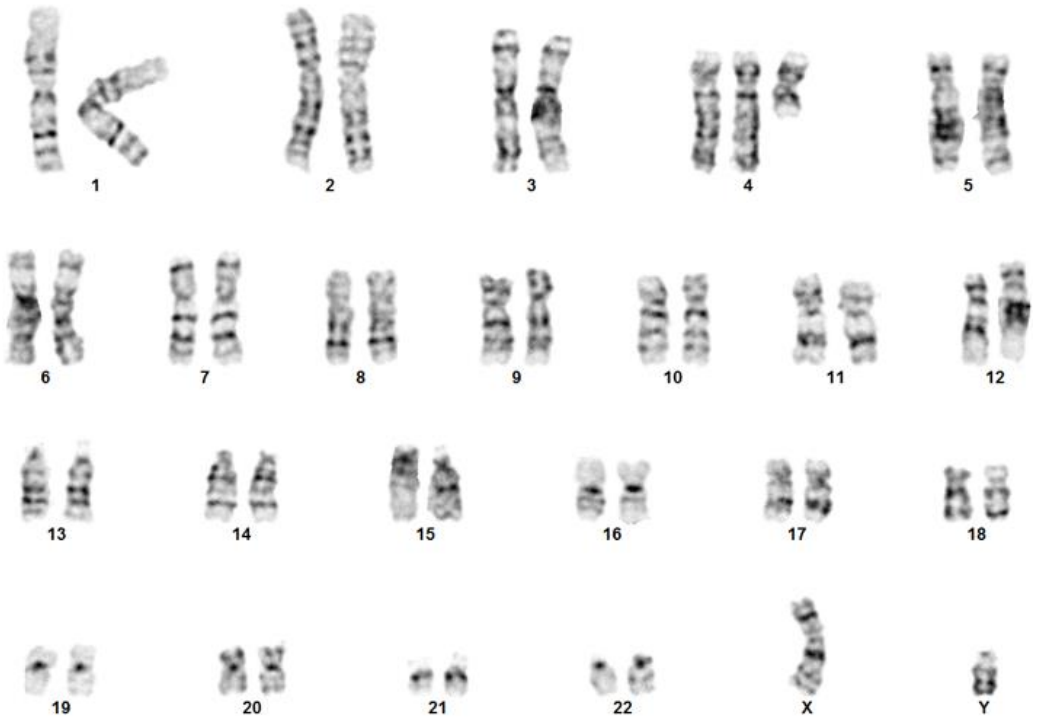

Figure 1: GTG karyotype demonstrating the gain of an isochromosome 4p

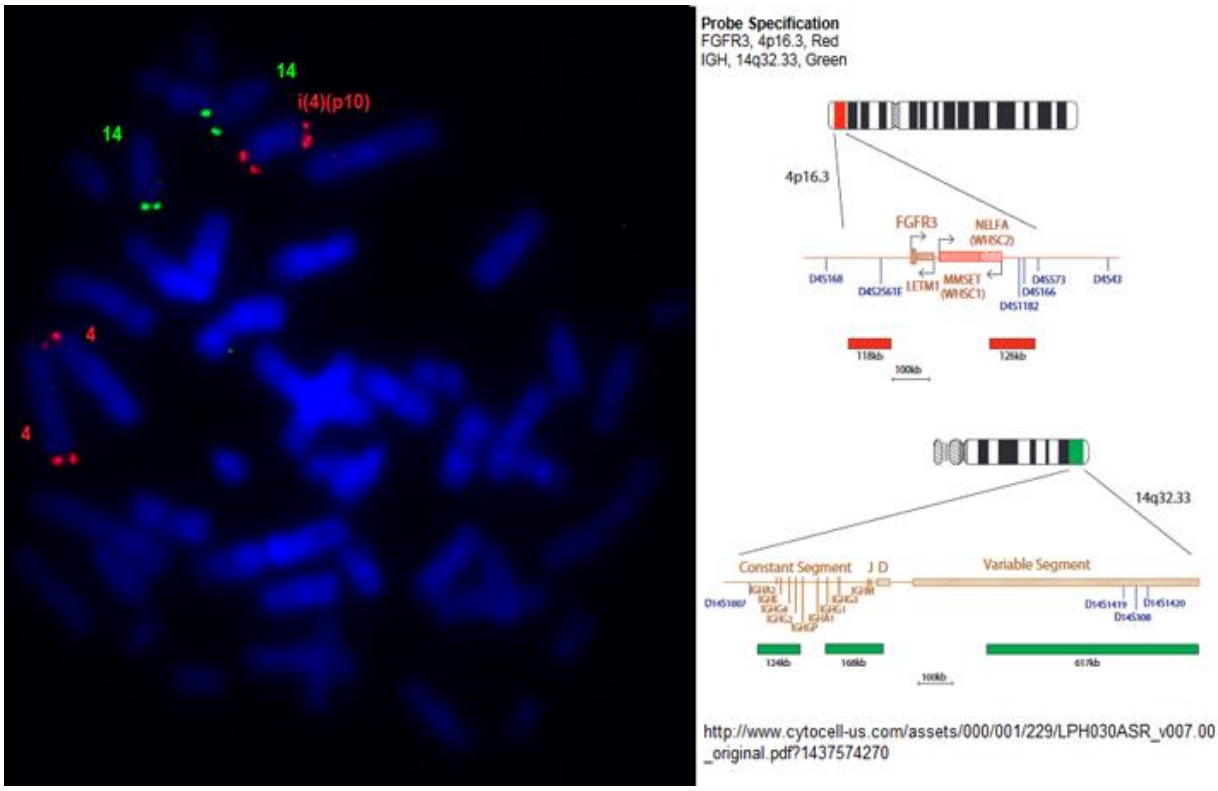

Figure 2: Metaphase plate FISH study using the FGFR3-IGH Dual Fusion probe (Cytocell LPH030) probe demonstrates the presence of 4 FGFR3 orange signals including two signals on the supernumerary isochromosome 4p.

Without additional prognostic data, this abnormality should be classified into the intermediate subgroup according to the ELN classification (Döhner et al., 2010).

The prevalent mechanism proposed to explain the oncogenic impact of the presence of a supernumerary i(4)(p10) is dosage effect of oncogenes located on the short arm of chromosome 4 (Chen et al., 1999; Soriani et al., 2010).

	Date	age	sex	FAB	CR	Follow-up
Case 1	1981	63	M	AML4	yes	6 months
Case 2	1995	46	M	AML4	yes	12 months
Case 3	1997	32	M	AML2	-	-
Case 4	1997	60	F	RAEB-T	-	-
Case 5	2010	43	M	AML4	yes	12 months
Case 6	2013	79	M	RAEB2	no	4 months
Case 7	2015	65	M	AML	yes	7 months

Table 1: cases of i(4)(p10) reported in myeloid neoplasms (M : male; F : Female; CR : complete remission)

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This article should be referenced as such:

Richebourg S, Fortin M, Gallagher G, Winstall E, Jacob S. A new case of adult Acute Myeloid Leukemia with isolated tetrasomy 4p. Atlas Genet Cytogenet Oncol Haematol. 2017; 21(8):310-312.