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Case Report Section

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Patient with t(4;12)(q11;p13) with therapy-related MDS and known history of stage II metastatic colorectal cancer

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Clinics

Age and sex
63 years old female patient.
Previous history
No preleukemia.
Previous malignancy: patient diagnosed with rectal cancer 8/03 treated with surgery and adjuvant chemo and radiation therapy. 9/2008 stage II isolated metastatic colorectal cancer involving the lung. Patient was treated with surgical resection and 5FU therapy and radiation. Patient is currently in remission. Has sibling with renal cell carcinoma.
No inborn condition of note
Organomegaly
No hepatomegaly, no splenomegaly, no enlarged lymph nodes, no central nervous system involvement.

Blood

WBC: 7.8X 10^9/l
HB: 11.2g/dl
Platelets: 176X 10^9/l
Blasts: 18% (18-29% bone marrow/ 4% peripheral blood).
Bone marrow : 28.3% segmented neutrophils, 10% eosinophils, 48.5% lymphocytes, 12.1% monocytes, 1.0% myelocytes, 2.0% metamyelocytes, and 3.0% atypical lymphocytes.

Cyto-Pathology Classification

Cytology
Trilineage dyspoiesis consistent with myelodysplasia.
Immunophenotype
CD7: 44.4%, CD13: 67.5%, CD34: 28.7%, CD117: 30.9%, HLA-DR: 38.5%, CD38: 71.4%.
Rearranged Ig Tcr
n/a
Pathology
Flow cytometric analysis of the specimen labeled bone marrow reveals a significant population of myeloblasts (approximately 29% of cells that could be studied in the sample) with most expressing abnormal CD7 (up to 1/3 is CD7 negative), CD13, CD33 (expression ranges from essentially undetectable up to moderately positive), CD34, CD38, CD45, CD117, and HLA-DR. Approximately half of the blast population is above the negativity threshold and shows dim to moderate intensity, CD123 expression. The remaining myeloid cells show minor abnormalities in antigen expression with respect to acquisition of full intensity CD13 and CD16. The change is less than typically occurs in well-defined MDS cases.
Electron microscopy
n/a

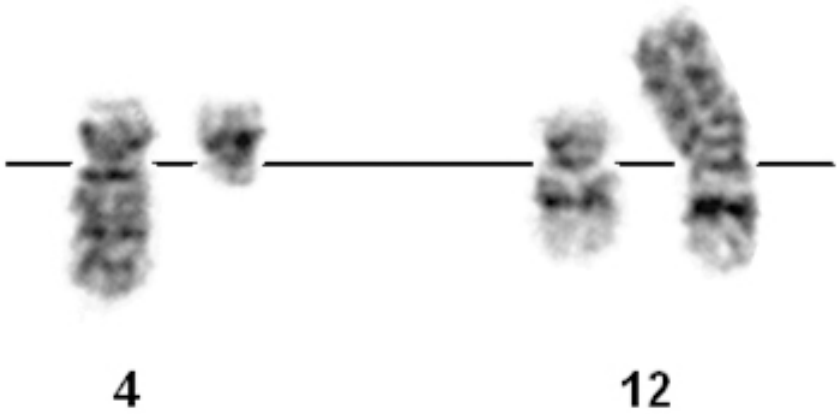

Fig. 1: Rare t(4;12)(q11;p13) found in a female patient with known history of metastatic colorectal cancer presenting with secondary high grade myelodysplastic syndrome.

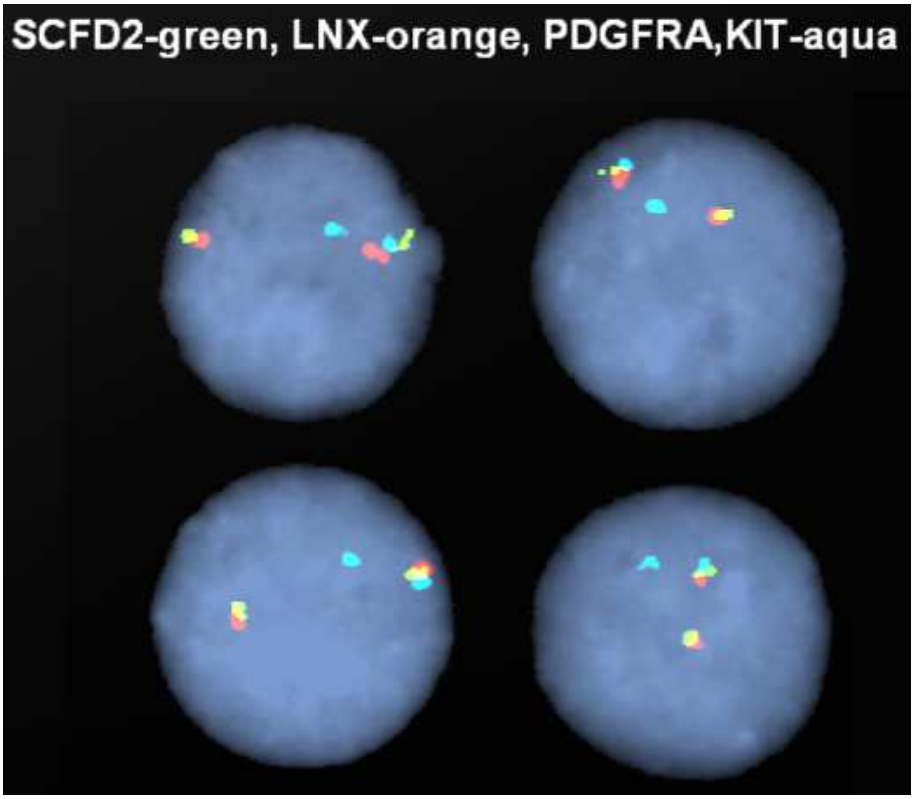

Fig. 2: Confirmation of 4q12 rearrangement using Vysis LSI 4q12 Tricolor Rearrangement Probe (Abbott).

Diagnosis

Flow cytometry- Acute myelogenous leukemia with ~29% myeloblasts. Morphology- therapy-related myelodysplasia with 18% blasts.

Survival

Date of diagnosis: 07-2010

Treatment: Patient is being treated with Vidasia, induction chemotherapy and salvage chemotherapy.

Complete remission: Refractory AML s/p chemotherapy.

Treatment related death: n/a

Relapse: n/a

Status: Alive

Last follow up: 03-2012

Survival: 20 months

Karyotype

Sample: Bone marrow

Culture time: 24 culture (unstimulated) and 48 hour culture (unstimulated).

Banding: GTW

Results

46,XX,t(4;12)(q11;p13)[20]

Karyotype at Relapse
n/a
Other molecular cytogenetics technics
Interphase FISH using Vysis LSI 4q12 Tricolor Rearrangement Probe (Abbott). Interphase FISH using LSI break apart probe for ETV6 (Abbott) was attempted on destained slides, no signals were detected.
Other molecular cytogenetics results
nuc ish(SCFD2,LNX,PDGFRA,KIT)x2 (SCDF2 con LNX sep PDGFRA,KITx1).

Other Molecular Studies

Technics:
OncoChip™ / CNE. Microarray analysis using a whole genome oligonucleotide array, which specifically targets genes, micro RNAs and specific genomic intervals with known or suspected relevance to cancer.
Results:
CNE Results. arr(1-22,X)x2 Normal Female.
Unclear findings. arr 7q34(141,693,456-141,719,136)x4,14q32.33(105,949,400-105,987,288)x0~1. Finding TCR and IGH rearrangements in the same clone is not unheard of as there is crosstalk. CNE hasn't been validated as a test for clonality.

Comments

Rare t(4;12)(q12;p13) found in a 63 year old female with history of stage II isolated metastatic colorectal cancer involving the lung. Results were confirmed by interphase FISH utilizing Vysis 4q12 Tricolor Rearrangement Probe (Abbott). Microarray studies were conducted using OncoChip™ whole genome oligonucleotide array, and yielded the following results: CNE result arr(1-22,X)x2 Normal Female, result with unclear clinical significance: arr 7q34(141,693,456-141,719,136)x4,14q32.33(105,949,400-105,987,288)x0~1.

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