

## Leukaemia Section

### Short Communication

## t(8;20)(p11;q13)

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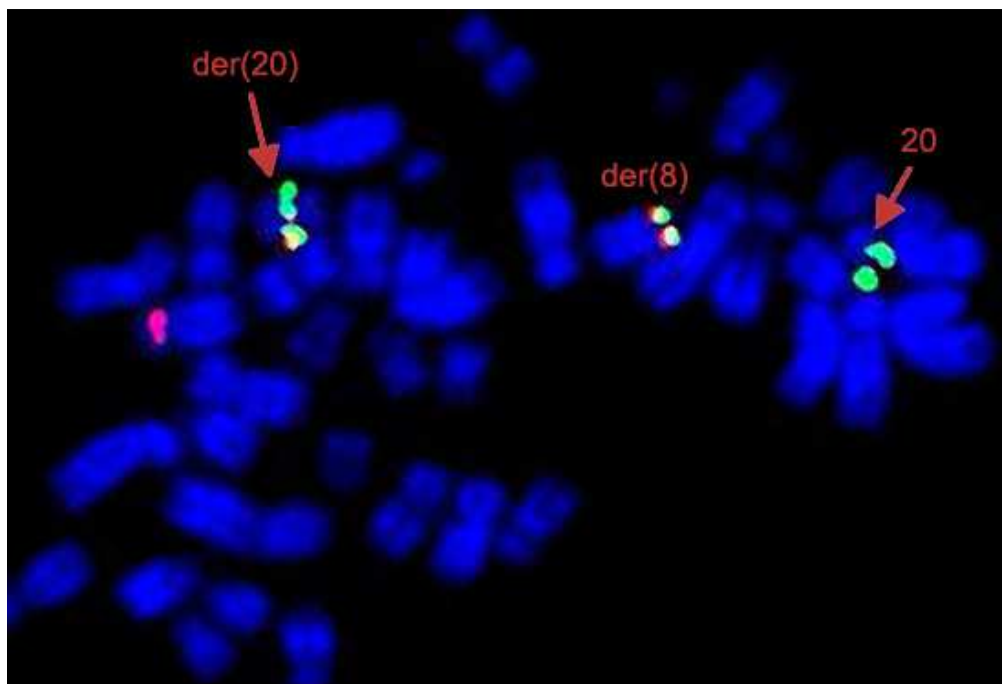
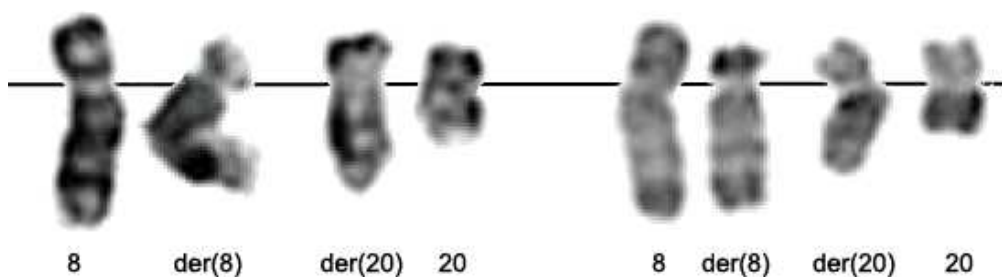
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### Identity



Top. GTG and R-banded partial karyotypes.

Bottom. FISH of metaphase chromosomes of t(8;20)(p11;q13) with digoxigenin-labeled RP11-313J18 (MYST3 at 8p11) and biotinylated RP11-1151C1 (5' and main part of NCOA3 region at 20q13) and RP11-122N8 (3' part of NCOA3 and SULF2 region at 20q13). Fused RP11-313J18 / RP11-1151C1+RP11-122N8 (red/green) signals are observed on der(8) and der(20) chromosomes. Courtesy of Christine Pérot and Marie-Joëlle Mozziconacci.

## Clinics and pathology

### Disease

Acute myeloid leukemia AML-M5 without features of erythrophagocytosis.

### Note

Only one case reported (a 75-year old woman). t(8;20) as a sole abnormality.

### Clinics

Splenomegaly, DIVC. Hyperleucocytosis, anemia and thrombopenia.

### Treatment

Hydroxyurea and low-dose cytosine-arabinoside.

### Evolution

Death 2 months after diagnosis. No remission obtained.

## Genes involved and proteins

### MYST3 (MYST histone acetyltransferase (monocytic leukemia) 3)

#### Location

8p11

#### Note

MYST3 is a histone acetyltransferase (HAT) belonging to the MYST family of HATs, that includes proteins involved in cell cycle regulation, chromatin remodeling and dosage compensation. MYST3 plays an important role during hematopoiesis with his transcriptional coregulator activity.

#### DNA/RNA

Breakpoint in intron 17.

#### Protein

MYST3 contains a LAP (Leukemia associated protein) zinc finger domain, a HAT domain (Histone acetyltransferase) and a acidic domain. 2004 amino acids; 225 kDa.

### NCOA3 (Nuclear Receptor Coactivator 3)

#### Location

20q13.1

#### Note

NCOA3 is a transcriptional coactivator that interacts with nuclear hormone receptors and with other transcription factors including TP52, NfκB and ER81. It has intrinsic histone acetyltransferase activity and recruits CREB Binding Protein (CBP)/p300 co-integrators into multisubunit coactivator complexes.

#### DNA/RNA

Breakpoint in exon 13 (Δ45 bp).

#### Protein

Member of the p160/steroid receptor coactivator family. 1424 amino acids; 155 kDa (130 kDa encoded by isoform b).

## Result of the chromosomal anomaly

### Hybrid gene

#### Note

Both MYST3-NCOA3 and NCOA3-MYST3 are expressed. Only the MYST3-NCOA3 fusion transcript has an open reading frame that may generate a functional chimeric protein.

### Fusion protein

#### Note

The CREB-interacting domain in NCOA3 (1046-1092aa) is conserved in the putative MYST3-NCOA3 fusion protein (at positions 1246-1292aa).

## References

Esteyries S, Perot C, Adelaide J, Imbert M, Lagarde A, Pautas C, Olschwang S, Birnbaum D, Chaffanet M, Mozziconacci MJ. NCOA3, a new fusion partner for MOZ/MYST3 in M5 acute myeloid leukemia. *Leukemia*. 2008 Mar;22(3):663-5

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