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in Oncology and Haematology

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Leukaemia Section
Mini Review

t(X;20)(q13;q13.3)

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Identity

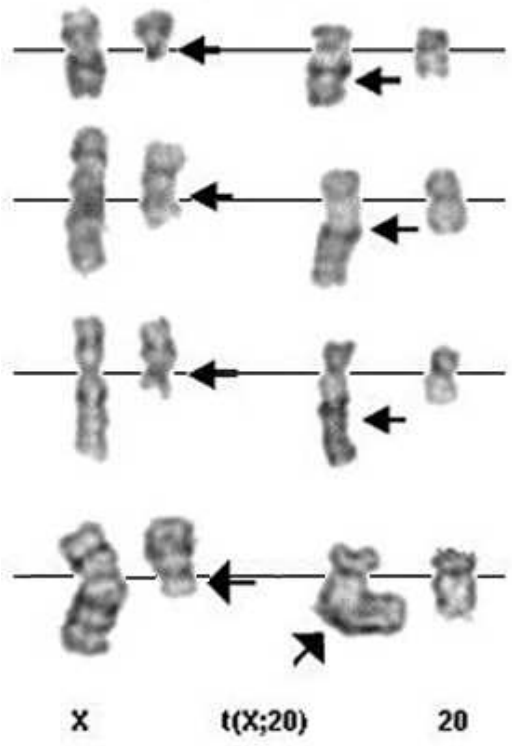

Fig. 1. Partial karyotypes of the translocation t(X;20)(q13;q13.3) for cases 1,4 (top to bottom; see case report section). Arrows indicate the derivatives 20 and X.

Clinics and pathology

Disease

Myelodysplastic syndromes (MDS): refractory anaemia with excess of blasts (RAEB-RAEBt), refractory anaemia (RA), MDS sideroblastic anemia and MDS pancytopenia and thrombocytopenia most often (5 cases); polycytemia vera --> acute myeloid leukemia (AML)-M1; myelofibrosis --> acute leukemia.

Epidemiology

Only 7 cases to date and they are exclusively female: 0 Male/7 Female; found in older patients (Median age 61 years; range: 57-86).

Clinics

Still poorly known.

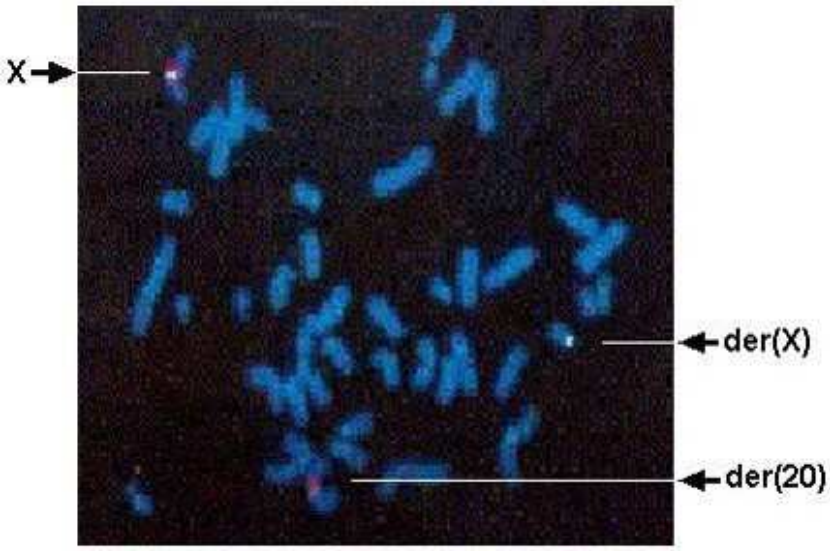

Fig 2: X-centromere probe DXZ1 (green) hybridized to the normal X and the derivative X (arrows). The androgen receptor (Xq12) AR (red) probe hybridized to derivative 20 and the normal X (arrows). The breakpoint on the X chromosome is proximal to AR. The karyotype is 46, X, t(X;20)(q13;q13.3).ish t(X;20)(q11.2q12;q13.3)(wcpX+,wcp20+,AR?;wcp20+, D20S180?,AR+,wcpX+). The revised breakpoints identified with FISH analysis are highlighted in bold.

Cytogenetics

Cytogenetics morphological

Sole abnormality in 5 MDS cases and accompanying changes +8, +9, del(13)(q21) and der(1;7)(q10;p10) in 2 cases that transformed to AML.

Cytogenetics molecular

By FISH the cytogenetic breakpoint was proximal to AR gene and hence the breakpoint on X chromosome is t(X;20)(q11.2q12;q13.3).

To be noted

Case Report

Translocation (X;20)(q13;q13.3): a nonrandom abnormality in four patients with myeloid disorders: case 1

Translocation (X;20)(q13;q13.3): a nonrandom abnormality in four patients with myeloid disorders: case 2

Translocation (X;20)(q13;q13.3): a nonrandom abnormality in four patients with myeloid disorders: case 3

Translocation (X;20)(q13;q13.3): a nonrandom abnormality in four patients with myeloid disorders: case 4

References

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