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Leukaemia Section
Short Communication

t(6;17)(p21;p13)

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Abstract

6p rearrangements in myeloid malignancies are characterized by heterogeneous breakpoints and chromosome abnormalities that involve various partner chromosomes. Balanced chromosome translocations involving 6p21 are infrequent, among them the t(6;17)(p21;p13 has been observed only in sporadic cases.

Keywords

Myeloid malignancies; 6p rearrangements; clonal evolution; t(6;17)(p21;p13).

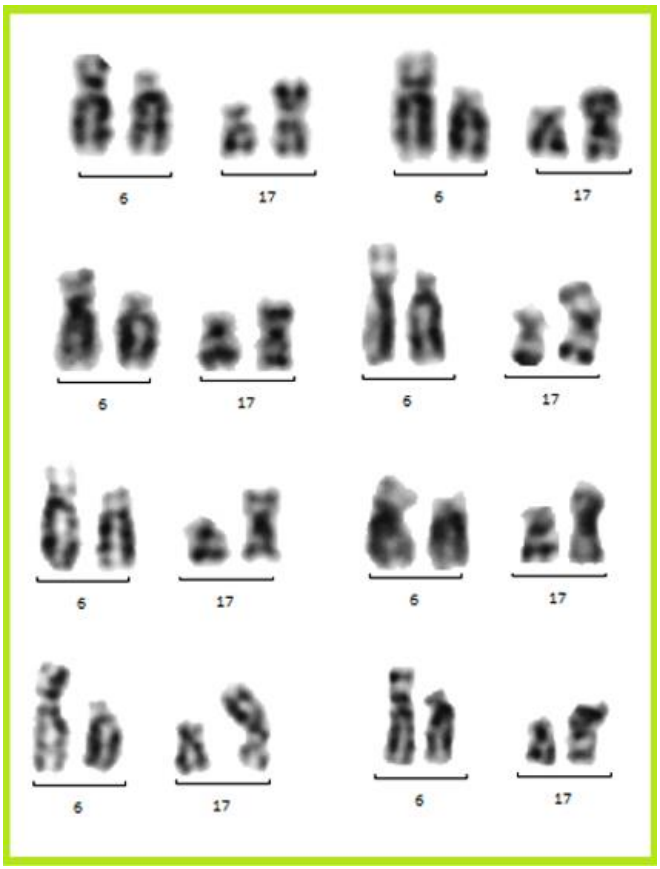

Figure 1. Partial karyotypes showing t(6;21)(p21;p13).

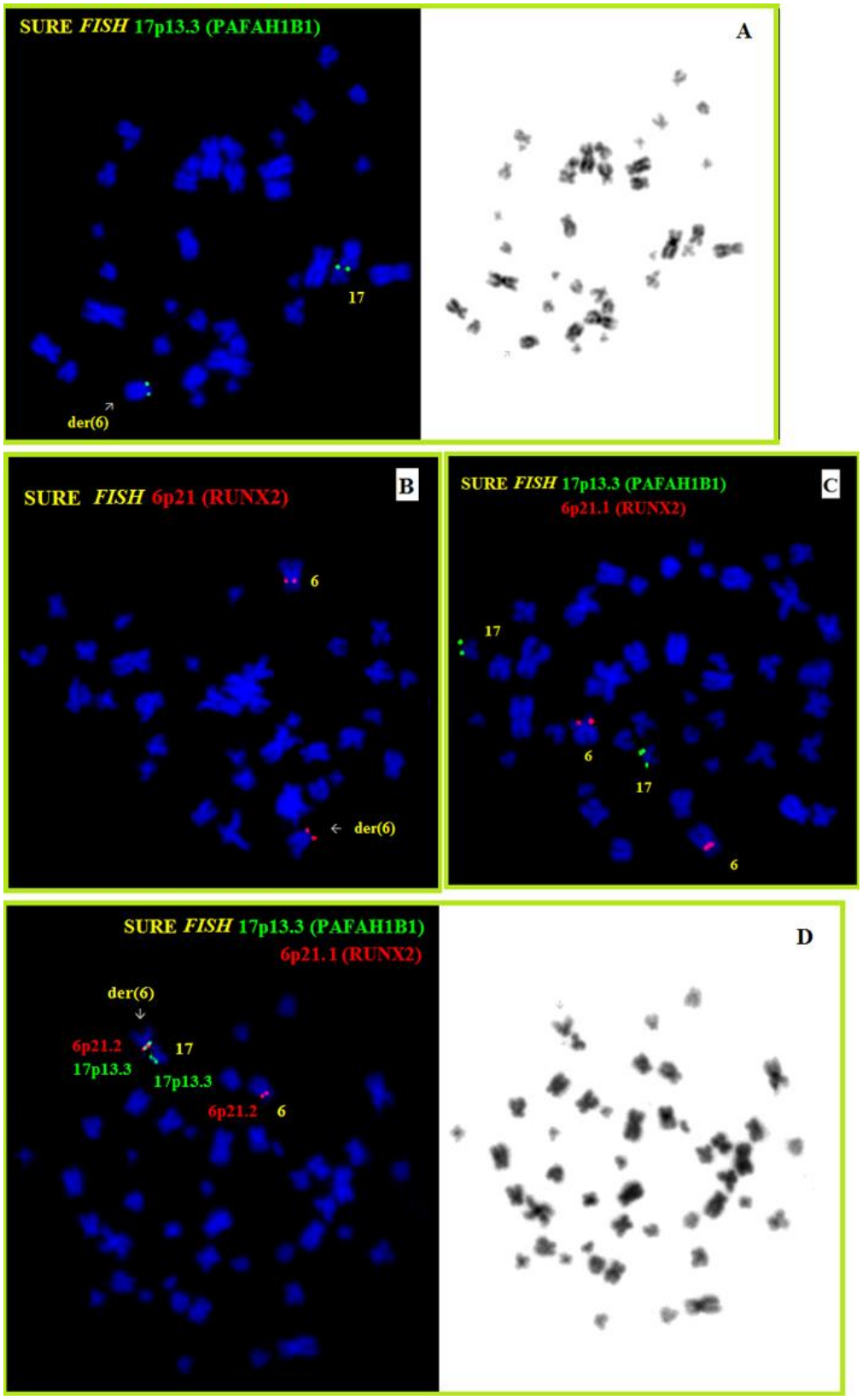

Figure 2. Hybridization with SureFISH PAFAH1B1 probe hybridizing to 17p13.3 showing translocation of 17p sequences to der(6) chromosome (green signal) (A). FISH with SureFISH RUNX2 probe located on 6p21.1 revealed signals on normal and der(6) chromosomes (B). Simultaneous hybridization with SureFISH PAFAH1B1 and RUNX2 probes showed normal signal pattern on metaphase without t(6;17)(p21;p13) (C) and cohybridization of PAFAH1B1 and RUNX2 probes on der(6) chromosome (red-green signal) (D).

Clinics and pathology

Disease

Myeloid malignancies

Phenotype/cell stem origin

1 acute myeloblastic leukemia with maturation (AML-M2) (La Starza et al., 2006), 1 myelodysplastic syndrome (MDS) that terminated in acute myeloid leukemia without maturation (AML-M1) and 1 AML with t(3;3)(q21;q26.2) (present cases, see the Case Reports t(6;17)(p21;p13) associated with t(3;3)(q21;q26.2) in AML and t(6;17)(p21;p13) and acquisition of the Philadelphia chromosome translocation with p190 BCR-ABL1 transcript during the course of myelodysplastic syndrome).
Note: an identical anomaly was also detected in 2 patients with aneurysmal bone cysts (Winnepenninckx et al., 2001; Althof et al., 2004).

Epidemiology

Only sporadic cases: 2 males aged 47 and 86 years and a 25-years old female.

Genetics

Note
Putative candidate genes at 6p21 include CCND3 at 6p21.1 and MHC complex, NOTCH4, BAK1, FANCE, ETV7, HMGA1, FKBP5 at 6p21.3 (La Starza et al., 2006).

Cytogenetics

Cytogenetics morphological

Found in association with +11 in AML-M2 and with +8 during MDS phase in the present patient in whom progression from MDS to AML was accompanied by an appearance of a new clone, t(9;22)(q34;q11) with the minor p190 BCR/ ABL1 transcript as an additional anomaly to initial chromosome abnormalities.
Found in a sideline in AML with t(3;3)(q21;q26.2) and monosomy 7.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis
The chromosomal translocation t(6;17)(p21;p13) is a rare anomaly that has been described in myeloid malignancies. Found in association with numerical chromosome anomalies such as +11, +8 and -7, therefore t(6;17)(p21;p13) is probably a secondary anomaly arising from a genetically unstable progenitor cell, acquiring subsequent genetic events.As these trisomies and monosomy 7 are known numerical aberrations in MDS and AML, it is likely that the occurrence of numerical anomalies may be a major pathogenetic event in these patients. Alternatively, it is possible that t(6;17)(p21;p13) was a primary anomaly associated with the early stage of disease that was replaced by a clone containing numerical anomalies during the course of a hematologic malignancy. The acquisition of t(9;22)(q34;q11) to initial anomalies in 1 patient indicates, that the Ph is certainly a secondary event that arose through multiple cytogenetic evolutions, the final event of which was the development of t(9;22)(q34;q11).

References

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