t(8;22)(p11;q11)

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Identity

Cytogenetics

Cytogenetics morphological
All three patients had a t(8;22)(p11;q11) identified by cytogenetics.

Genes involved and proteins

FGFR1 (fibroblast growth factor receptor 1)
Location 8p11
Protein Receptor tyrosine kinase for FGF ligands, also involved in the t(6;8)(q27;p12), t(8;9)(p12;q32-34) and t(8;13)(p12;q12).

BCR (breakpoint cluster region)
Location 22q11
Protein Normal function unclear; fuses to ABL in CML.

Result of the chromosomal anomaly

Hybrid gene
Description 5' BCR - 3' FGFR1
Transcript All three patients has BCR exon 4 fused to FGFR1 exon 9.
Detection RT-PCR or FISH.

Clinics and pathology

Disease
Described in only 3 cases, all of whom had a CML-like (chronic myelogenous leukemia-like) disease. Two had also had an excess of B-lymphocytes in the marrow.

Phenotype/cell stem origin
Involvement of both myeloid and lymphoid cells, so likely to be a stem cell disorder.

Evolution
At the time of writing, one patient had undergone myeloid transformation and a second patient underwent lymphoid transformation followed shortly by myeloid transformation. In common with other patients with FGFR1 fusion genes, the disease therefore appears to be aggressive.

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**Fusion protein**

**Description**
Contains the coiled-coil domain of BCR, the BCR Y177 Grb2 binding site and the entire tyrosine kinase domain of FGFR1.

**Oncogenesis**
Analogous to BCR-ABL: constitutive activation of FGFR1 tyrosine kinase. Transforms Ba/F3 cells to IL-3 independence. Transformed cells are not affected by imatinib (Gleevec/Glivec/STI571) but are inhibited by compounds with anti-FGFR activity.

**References**


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