Leukaemia Section
Short Communication

\[ t(8;17)(p12;q23) \]

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Clinics and pathology

Disease
Myeloproliferative disease (MPD)

Phenotype/cell stem origin
The disease was characterized by thrombocytopenia due to decreased and dysplastic megakaryocytes, an elevated number of monocytes, eosinophils and basophils.

Epidemiology
Only one case to date, a 74 year old female patient.

Clinics
The patient remained in a stable condition for 2 years and finally died due to treatment-resistant disease progression.

Genes involved and proteins

\textit{FGFR1}

Location
8p12

Protein
FGF receptor with tyrosine kinase activity; binding of ligand induces receptor dimerization, auto-phosphorylation and signal transduction.

\textit{MYO18A}

Location
17q11.2

Protein
Member of the myosin family. MYO18A, also called MysPDZ, contains from N-term a KE-rich domain, an ATP-insensitive actin-binding site (able to link to actin), a PDZ domain, a myosin head, an IQ motif, a coiled-coil domain, and a globular domain in C-term. This myosin might stably cross link actin filaments for high order organization of the actin cytoskeleton (Isogawa et al., 2005).

Result of the chromosomal anomaly

Hybrid gene

Note
The two genes are transcribed in opposite direction, and an inversion is necessary to produce the hybrid gene and protein.

Description
5' MYO18A - 3' FGFR1; fusion of MYO18A exon 32 to FGFR1 exon 9.

Fusion protein

Description
1691 amino acids from MYO18A, including the PDZ protein-protein interaction domain, the myosin head domain, and a coiled-coil domain, fused to 394 amino acids from FGFR1, including part of its juxta membrane domain, and its tyrosine kinase domain; may induce dimerization, and constitutive signal transduction.

References


This article should be referenced as such: