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Leukaemia Section

Short Communication

t(1;5)(q22;q33)

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Clinics and pathology

Disease

Myeloproliferative disorders (MPD) with eosinophilia (or chronic eosinophilic leukemia (CEL)) and B-cell acute lymphoblastic leukaemia (ALL).

Epidemiology

4 cases of MPD with eosinophilia, and 1 case of ALL; because the phenotypes are different, it may be that genes involved in this/these disease(s) are not similar; PDE4DIP and PDFRGB were found involved in MPD with eosinophilia (see below).

Clinics

MPD cases were found in 3 infants (aged 5, 7, and 11 months), and one young adult, aged 21 years; sex ratio was 1/1. The ALL case was a 13 year old boy.

Prognosis

One MPD case died 9 mths after diagnosis, another one was alive at 14 months+, one was alive and well 7 years after diagnosis with IFN therapy, and one was found resistant to various treatments, including IFN therapy, until -because PDGFRB was found to be involved in the disease- imatinib was started and remission obtained. The ALL case experienced difficult remission, relapse, BM transplantation, CNS relapse; the parents finally refused further therapy and the patient died.

Cytogenetics

Additional anomalies

Sole anomaly in each case.

Genes involved and Proteins

PDE4DIP (phosphodiesterase 4D interacting protein)

Note: 1q22

Protein

PDE4DIP codes for a protein called myomegalin; interacts with the cyclic nucleotide phosphodiesterase PDE4D; there ar at least 2 isoforms of myomegalin: KIAA0454 isoform and KIAA0477 isoform.

PDGFRB

Location: 5q33

Protein

PDGFRB is the receptor for PDGFB (platelet-derived growth factor-b); Ig like, transmembrane and tyrosine kinase domains; membrane tyrosine kinase; can homodimerize.

Results of the chromosomal anomaly

Hybrid gene

Description

5' PDE4DIP - 3' PDGFRB; PDE4DIP (KIAA0477 isoform) fuses in frame PDGFRB exon 11.

Transcript

The reciprocal PDGFRB-PDE4DIP is not expressed.

Fusion protein

Description

The first 905 amino acids of PDE4DIP, including the coiled-coil domains are fused to the transmembrane and the tyrosine kinase domains of PDGFRB.

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