Leukaemia Section

Short Communication

t(4;11)(q35;q23)
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Clinics and pathology

Disease
Acute myeloid leukemia (AML)

Epidemiology
Only one case to date, a 3-month-old girl with M5 AML.

Prognosis
No data.

Genes involved and Proteins

SORBS2
Location: 4q35.1
Note: This gene is better known under the name ArgBP2.

Protein
From N-term to C-term, the protein contains a sorbin homology (SoHo) domain and three SH3 domains. Adaptor protein. Binds signaling proteins like ABL1 and ABL2, AKT and PAK1, and various cytoskeletal proteins, including vinculin, alpha actinin, spectrin. Role in cytoskeleton organization, lipid rafts, and signaling.

MLL
Location: 11q23

Protein
Transcription regulator (yin/yang?), regulates, among others, HOX genes expression ---> Hematopoiesis and embryogenesis regulation.

Results of the chromosomal anomaly

Hybrid gene
Description
5' MLL - 3' SORBS2

Transcript
Fusion transcript of MLL exon 7 to SORBS2 exon 19; the reciprocal was not detected.

Fusion protein
Description
N-term AT hook and methyl transferase domains from MLL, fused to the third and last C-term SH3 domain of SORBS2.

Expression localisation
The MLL-SORBS2 hybrid localizes to the nucleus with a punctate pattern characteristic of MLL chimeric proteins.

References


This article should be referenced as such: