

Leukaemia Section

Short Communication

t(4;11)(q35;q23)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France

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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.

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