t(1;9)(q24;q34)
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Identity

R-banded karyotype showing the t(1;9)(q24;q34) translocation.

Disease
B-cell acute lymphoblastic leukemia.

Epidemiology
Only 1 case to date, a 11-year-old boy.

Prognosis
Complete remission was obtained and a bone marrow transplantation was performed.

Cytogenetics

Cytogenetics, molecular
LSI bcr/abl dual extra-signal (ES) color probe (Abbott, Rungis, France) and BAC Probes.

Probes
RP11-83J21 (chromosome 9) and RP11-232M22, RP11-928F1, RP11-138P14, RP11-652E14, RP11-64D9 (chromosome 1).
All the probes that were used to find the breakpoint on der(1).

Genes involved and Proteins

ABL1 (Abelson Murine Leukemia Viral Oncogene Homolog 1)

Location: 9q34
DNA / RNA
The ABL gene is approximately 225 kb in size and is expressed as a 7-kb mRNA transcript, with alternatively spliced first exons, exons 1b and 1a, respectively, spliced to the common exons 2-11. Exon 1b is approximately 200 kb 5-prime of exon 1a.

Protein
The 145-kD ABL protein is classified as a nonreceptor tyrosine kinase. When the N-terminal region of the ABL protein is encoded by exon 1a, the protein is believed to be localized in the nucleus, while when encoded by exon 1b, the resulting N-terminal glycine would be myristylated and thus postulated to direct that protein to the plasma membrane.

RCSD1 (RCSD Domain-Containing Protein 1)

Location: 1q24
DNA / RNA
Eyers et al. (2005) cloned for the first time the human RCSD1, which they called CAPZIP. A 416-amino acid protein was deduced and they calculated a molecular mass of 44.5 kD. Northern blot analysis resulted in a major 3.4-kb transcript and a minor 7-kb transcript that is highly expressed in skeletal muscle and weakly in cardiac muscle. CAPZIP is detected in several lymphoid organs, including spleen, thymus, peripheral blood leukocytes, lymph node, and bone marrow.
Protein

Eyers et al. (2005) found many properties of rabbit Capzip. It interacted specifically with the F-actin capping protein CapZ. This protein was phosphorylated by: MAPKAPK2 and SAPK3 (MAPK12), on ser108 by SAPK3 and SAPK4 (MAPK13) and on ser68, ser83, and ser216 by JNK1 alpha-1 (MAPK8) in vitro. This team also found that stress induced by hyperosmotic shock and anisomycin, a protein synthesis inhibitor, stimulated the phosphorylation of CAPZIP in human
cell lines and induced the dissociation of CAPZIP from CAPZ in Jurkat human T cells. This phenomenon may regulate the ability of CapZ to remodel actin filament.

### Results of the chromosomal anomaly

#### Hybrid gene

**Description**
The 3' region of ABL1 is translocated on the 5' region of RCSD1 on the der(1) and the 3' region of RCSD1 is translocated on the 5' region of ABL1 on der(9).

**Detection protocol**
FISH detection.

#### Fusion protein

**Description**
The fusion gene is predicted to encode an aberrant tyrosine kinase.

#### Oncogenesis

The RCSD1 gene, which codes a protein kinase substrate, CapZIP (CapZ-interacting protein), is found in immune cells, splenocytes and muscle. It is possible that the interaction between CapZIP and CapZ affects the cell ability to remodel actin filament assembly. CapZIP is phosphorylated when cells are exposed to various cellular stresses, which activate the kinase cascade. The interaction between CapZIP and CapZ would be lost when CapZIP is phosphorylated. So, RCSD1 would be involved in the remodeling of the actin cytoskeleton, which is an important step in mitosis. The probable formation of the ABL1-RCSD1 fusion gene could result in an alteration of the cellular function by affecting the cytoskeleton regulation, which could be an important step in leukemogenesis.

### References


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