

del(11)(p12p13)

Clinics and Pathology

Disease [T-cell acute lymphoblastic leukemia \(T-ALL\)](#).

Epidemiology About 5% of T-ALL patients.

Prognosis Currently, no relation between the cryptic deletion, del(11)(p12p13), and prognosis could be established. This could be due to the limited patient numbers in the study.

Genetics

The cryptic deletion, del(11)(p12p13) was identified using microarray-based comparative genome hybridisation (array-CGH). The deleted region is about 3 Mb in size and the telomeric breakpoint of these deletions is situated in or near the LMO2 oncogene. Variances in the centromeric breakpoints is detected.

Cytogenetics

Variants One of the T-ALL patients showed a cryptic deletion, del(11)(p12p13), that did not target the LMO2 oncogene. Indeed, this case showed no ectopic LMO2 expression. Therefore, this genomic region could potentially contain a tumor suppressor gene that also contributes to T-ALL pathogenesis.

Genes involved and Proteins

Gene Name [RBTN2/LMO2](#)

Location 11p13

Protein LMO2 encodes a protein that participates in the transcription factor complex, which includes [E2A](#), [TAL1](#), [GATA1](#), and [LDB1](#) in erythroid cells. Within this transcription complex, LMO2 mediates the protein-protein interactions by recruiting LDB1, whereas TAL1, GATA1, and E2A regulate the binding to specific DNA target sites. This complex regulates the expression of several genes in various cellular backgrounds including [C-KIT](#), [EKLF](#), and [RALDH](#). In normal T-cell development, LMO2 is expressed in immature CD4/CD8 double-negative thymocytes, and is down-regulated during T-cell maturation.

Result of the chromosomal anomaly

Hybrid gene

Note Ectopic expression of the LMO2 oncogene due to the removal of a negative regulatory element situated upstream of the LMO2 gene, leading to activation of the proximal LMO2 promoter.
In one T-ALL case, this recurrent deletion resulted in a RAG2-LMO2 fusion gene, bringing the LMO2 gene under the control of RAG2 promoter sequences. However, it was shown that promoter substitution was not the main activational mechanism as none of the other del(11)(p12p13) positive cases showed a similar RAG2-LMO2 fusion gene. In addition, RQ-PCR analysis revealed that the expression of the RAG2-LMO2 fusion is much lower than the wildtype LMO2 expression from the proximal LMO2 gene promoter.

External links

Other database [del\(11\)\(p12p13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Bibliography

The cryptic chromosomal deletion del(11)(p12p13) as a new activation mechanism of LMO2 in pediatric T-cell acute lymphoblastic leukemia.

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