Leukaemia Section
Short Communication

t(3;9)(p13;q34.1) FOXP1/ABL1

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Abstract
Review on t(3;9)(p13;q34.1) FOXP1/ABL1, with data on clinics, and the genes involved.

Keywords
chromosome 3; chromosome 9; FOXP1; ABL1; B Cell Acute Lymphoblastic Leukemia; Follicular Lymphoma

Clinics and pathology

Disease
Pre-B Cell Acute Lymphoblastic Leukemia/Lymphoma (ALL) and Follicular Lymphoma (Ernst et al., 2011; Koduru et al., 1997).

Note
The FOXP1/ABL1 involvement was ascertained only in the B-ALL case. t(3;9) was found in a subclone and against the background of a complex karyotype and TP53 gene mutation in the follicular lymphoma case.

Phenotype/cell stem origin
Pre-B cell.

Epidemiology
1 ALL case reported to date: a 16 yo female patient, and 1 follicular lymphoma case reported to date: a 52 yo male patient.

Cytology
High leukocytosis (>50,000 x 10⁹/L) at diagnosis.

Treatment
For ALL: Standard COALL (German Cooperative Study Group) protocol for high-risk ALL followed by Allogeneic BMT; may be sensitive to treatment with first or 2nd generation tyrosine kinase inhibitors.

Prognosis
Yet unknown; ALL patient was reported in remission 9 years post diagnosis, but after paternal origin haplo-identical BMT.

Cytogenetics

Probes
ABL1 should show split signal using the standard DCDF BCR/ABL1 construct.

Genes involved and proteins

FOX1 (Forkhead box P1)

Location
3p13

Note
Member of forkhead box (FOX) subfamily P; transcription factor. These proteins play a role in cell- and tissue-specific gene transcription regulation.

DNA/RNA
Gene is 176,228 bp with 16 exons; transcribed from the - strand; coding region is 171,437 bp with 14 exons.

Protein
At least 12 protein isoforms produced; dimerizes with FOXP2 and FOXP4 using the leucine-zipper domain which is required for DNA binding capability. Protein locates to the nucleus.

Germinal mutations
Germline mutations of FOXP1 are associated with autosomal dominant intellectual disability with language impairment, with or without autistic features (MIM phenotype 613670).

**ABL1 (v- abl Abelson murine leukemia viral oncogene homolog 1)**

**Location**  
9q34.12

**DNA/RNA**  
Expressed as either 6- or 7-Kb transcript.

**Protein**  
Tyrosine kinase; located in either the nucleus (shorter transcript) or cytoplasm (longer transcript) depending on which splice variant is produced (Chissoe et al., 1995).

**Result of the chromosomal anomaly**

**Hybrid gene**

**Note**  
In the single patient characterized, the in-frame fusion was confirmed between FOXP1 exon 19 (ENST00000318789) and ABL1 exon 4 (ENST00000318560) (Ernst et al., 2011).

**Description**  
ABL1 exon 4 fused with FOXP1 alternative RNA isoform (NM_032682).

**Detection**

RT-PCR

**References**


This article should be referenced as such:

Sanford Biggerstaff JA. t(3;9)(p13;q34.1) FOXP1/ABL1. Atlas Genet Cytogenet Oncol Haematol. 2017; 21(7):263-264.