t(9;15)(p13;q24) PAX5/GOLGA6A

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Abstract
Review on t(9;15)(p13;q24) PAX5/GOLGA6A, with data on clinics, and the genes implicated.

Clinics and pathology

Disease
B-cell acute lymphoblastic leukemia (B-ALL)
Note
Was not taken into account in this review a case of diffuse large B-cell lymphoma in a 51 year-old male patient (Morrison et al., 1994).

Epidemiology
One case to date, a 21-year-old male patient with a CD10+ (B-II, common) ALL (Coyaud et al., 2010).

Prognosis
No data.

Cytogenetics

Cytogenetics morphological
There were additional numerical anomalies (+5, +21).

Genes involved and proteins

PAX5
Location
9p13.2

Protein
391 amino acids; from N-term to C-term, PAX5 contains: a paired domain (aa: 16-142); an octapeptide (aa: 179-186); a partial homeodomain (aa: 228-254); a transactivation domain (aa: 304-359); and an inhibitory domain (aa: 359-391). Lineage-specific transcription factor; recognizes the consensus recognition sequence GNCCANTGAAGCGTGAC, where N is any nucleotide. Involved in B-cell differentiation. Entry of common lymphoid progenitors into the B cell lineage depends on E2A, EBF1, and PAX5; activates B-cell specific genes and repress genes involved in other lineage commitments. Activates the surface cell receptor CD19 and repress FLT3. Pax5 physically interacts with the RAG1/RAG2 complex, and removes the inhibitory signal of the lysine-9-methylated histone H3, and induces V-to-DJ rearrangements. Genes repressed by PAX5 expression in early B cells are restored in their function in mature B cells and plasma cells, and PAX5 repressed (Fuxa et al., 2004; Johnson et al., 2004; Zhang et al., 2006; Cobaleda et al., 2007; Medvedovic et al., 2011).

GOLGA6A
Location
15q24.1

Protein
693 amino acids (aa); contains a coiled coil domain (aa 14-611), with 11 leucine zipper motifs (aa 360-430).
Member of the golgin family of proteins, localizes to the Golgi apparatus. Its function is poorly known (Gilles et al., 2000).

**Result of the chromosomal anomaly**

**Hybrid gene**

*Description*
Fusion of PAX5 exon 6 to GOLGA6A exon 3.

**Fusion protein**

*Description*
887 amino acids. The predicted fusion protein contains the DNA binding paired domain, the octapeptide, and the homeodomain of PAX5 and part of the coiled coil domain of GOLGA6A.

**References**


This article should be referenced as such: