TTL (twelve-thirteen translocation leukemia)
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Identity

Other names: FLJ21437; LOC646982; TTL/TEL; TTL-T; TTL-B1; TTL-B2
Location: 13q14.11
Note: Not to be confused with: TTL : tubulin tyrosine ligase (2q13), nor with ‘transthretin-like (TTL) gene family’, a family to which belongs TTR (transthretin, 18q12).

DNA/RNA

Description
Start at 39,822,377 bp from pter; the gene spans 119,929 bases on minus strand.

Transcription
Three splicing forms, namely: TTL-T, TTL-B1 and -B2. TTL-T is 2090 bp long and composed of exons 1-8. The longest open-reading frame contains exons 4, 5, and part of exon 6; it encodes a 133 amino acids peptide. TTL-B1 transcript is 3450 bp long and is composed of exons 4, 5, and part of exon 9. TTL-B2, 3588 bp long is composed of exons 4, 5, and part of exon 8a.

Protein

Note: This gene/protein remains poorly known: there has been no study on it since the princeps paper by Qiao et al (2003).

Expression
Ubiquitous expression (lung, liver, spleen, thymus, and bone marrow); major expression in brain and testis.

Homology
TTL has no homology to known genes.

Implicated in

\[ t(12;13)(p13;q14) \text{ in B-cell acute lymphoblastic leukaemia (B-ALL)} \rightarrow ETV6/TTL \]

Note: Only one case to date.

Hybrid/Mutated Gene
Both reciprocal transcripts, TTL/ETV6 and ETV6/TTL, were detected. ETV6/TTL fusion transcript.

The other transcript, TTL/ETV6, comprises 5’ TTL exons 1 to 5 or to 8a, fused to ETV6 from exon 2. The predicted 530 amino acids fusion protein consists mostly of ETV6 with both HLH and ETS domains, and could have modified transcriptional activities. On the other hand, a loss of function of ETV6 and/or of TTL could play the critical role in leukemogenesis.

References

This article should be referenced as such: