Leukaemia Section
Short Communication

t(2;11)(q33;q23) KMT2A/ABI2

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Abstract
Review on t(2;11)(q33;q23) KMT2A/ABI2, with data on clinics, and the genes implicated.

Clinics and pathology

Disease
M5 acute non lymphocytic leukemia (AML)

Clinics
poorly known: one case, 5-months old girl with 46,XX,t(2;11)(q33;q23)

Genes involved and proteins

KMT2A
Location
11q23
DNA/RNA
21 exons, spanning about 100kb; 13-17 kb mRNA.
Protein
431 kDa; contains two DNA binding motifs (an AT hook and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation.

ABI2
Location
2q33
DNA/RNA
16 exons, spanning about 104kb, about 22kb mRNA.
Protein
513 amino acids containing protein, different isoforms described, contains an Abl-interactor homeodomain homologous region (HHR), SRC homology 3 (SH3) domains and proline rich stretches.
Functions as inhibitor of c-Abl.

Result of the chromosomal anomaly

Hybrid gene
Transcript
3 different transcripts were detected, see above.
Detection
RT-PCR according to reference.
t(2;11)(q33;q23) KMT2A/ABI2 fusion product. DNA sequence is shown as derived from a long distance inverse PCR experiment. Shown in black is the sequence from MLL intron 8, exon 9 (bold) and intron 9, respectively, in blue two inserted nucleotides, in red the sequence from ABI2 intron 4. The schematic overview of transcripts shows the different splice variants that were present. cDNA sequence is shown as derived from Reverse Transcription-PCR using an exon 8 specific MLL and an exon 6 specific ABI2 primer. The gel picture shows multiple bands that were derived with these primers and were confirmed by cloning.

References

This article should be referenced as such: