

Leukaemia Section

Short Communication

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

Eva A Coenen, C Michel Zwaan, Marry M van den Heuvel-Eibrink, Claus Meyer, Rolf Marschalek, Ursula Creutzig, Rob Pieters, Jutta Bradtke

Department of Pediatric Oncology and Hematology, Erasmus MC, Sophia Children's Hospital, Rotterdam, The Netherlands; Institute of Pharmaceutical Biology, ZAFES, Diagnostic Center of Acute Leukemias (DCAL), Frankfurt, Germany; AML-BFM Study Group, Hannover, Germany; Department of Pediatric Hematology and Oncology, Justus-Liebig-University, Giessen, Germany; m.vandenheuvel@erasmusmc.nl

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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.

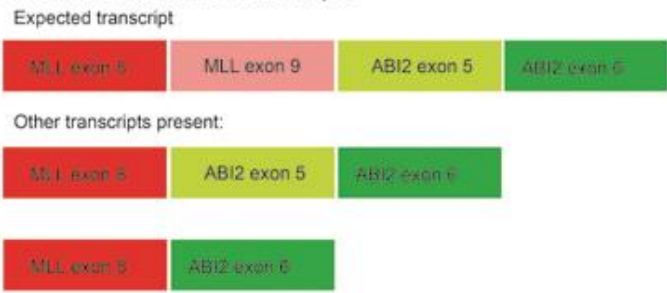
Detection

RT-PCR according to reference.

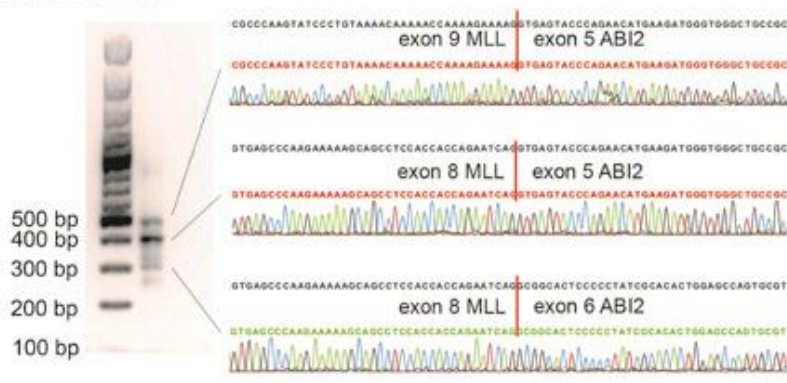
DNA sequence

TGAAGTCTTCAGTTCAAGAAAATCAGCTCTCTTTCTAACTATTATGTTTAATAATA
AAGAAACAGAAACAAAAAACAGTTAAATTGGAGGTATTGTTTTAATTCCTGT
TCGAAGCCTAGAGTTAAATAGTTTTTTTTTTTTTTCTAATGGCCCTTCTTCA
CAGGTAGCAGTCAGTACTAAAGTAGTCGTTGCCAGCATCTGACTGCAATTTATT
CTGAATTTTTAG**GTCCAGAGCAGAGCAAACAGAAAAAGTGGCTCCCCG**
CCCAAGTATCCCTGTAAAAACAAAAACAAAAGAAAAAGGTGAGGAGAGATT
TGTTTCTCTGCCATTTCTCAGGGATGATTCTATTTTGTAGGGAAAAGCCTTATC
CTTGACTTCTATGTAGATGGCAGTGGAAATTTCTTAAAAATTAAGAACTTCAAGTT
TAGGCTTTTAGCTGGGCACGGTGGCTCACGCTGGTAATCCCAACACTTAGTGA
GGCTGAGGTGGGAGGATTGCTTGAGGCCAGCAGTTCAAGACCAGCCTGGGCA
ACATAGCAAGACCTGTCTTTATTTAAACAAAAAAGAAAGAAAGAAGAAGA
AGAAGT•TA•CAGAAGATACTGCAATTTAAACAAAAACCTTGCATACCAAAA
CTTAAAACAAATATAAGCTGGAAAGCTCTTTCAAATATTTAGCTATCCCATGT
ATAGAATAAAATCTCTCTTAAATTCCTGCTCTTTTCTTCTGTTTCTCAGCA
ATTGATGTAGTTTTAGTGTGGTTCACTTCTCCCTCATCACTTAAAGTAACTTTG
GGTTTTATGTTGTTAAGGTTGAGCTTAAAGGACTTTGGTTACCACTTTAAAAA
ATCTTCTGGTTTCAAGAAATTAAGATATGCAGTTTTAAAGCTATTCAATTTGTC
ATATTACTCCTAAAATTTCCCAAATGTGGATTGATTTTTTAAAAAACTTTTT
CTGGCATTTCATTTAAGATAACAAGTAGTATACCCTCCGCTTATTAATGAGAGG
GAGAGCAAGGATAAGTTAATCTTACTTACTTGATAATGTTTGTTTTATTGTT
TAAAAGCAACAACAAAAGGATTACAGA

Schematic overview of transcripts



cDNA sequence



MLL-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

Coenen EA, Zwaan CM, Meyer C, Marschalek R, Creutzig U, Pieters R, Bradtke J, van den Heuvel-Eibrink MM. Abl-interactor 2 (ABI2): a novel MLL translocation partner in acute myeloid leukemia. *Leuk Res.* 2012 May;36(5):e113-5

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