t(3;11)(q13.13;q23) MLL/KIAA1524

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Clinics and pathology

Disease

M5 acute non lymphocytic leukemia (AML)

Clinics

Poorly known: one case, a 4-months old girl with 46,XX,t(3:11)(q12~13;q23).

A. DNA sequence as derived from a Long distance inverse-PCR experiment. Shown in black is the sequence from MLL intron 9, exon 10 (Bold) and intron 10 respectively, in blue two inserted nucleotides, in red the sequence from KIAA1524 intron 16, exon 17 (Bold) and intron 17 respectively. (figure adapted from reference).
Genes involved and proteins

**MLL**

**Location**
11q23

**DNA/RNA**
21 exons, spanning about 100 kb; 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.

**KIAA1524**

**Location**
3q13.13

**Note**
Alias: CIP2A.

**DNA/RNA**
21 exons, spanning about 40 kb; 3-4 kb mRNA.

MLL-KIAA1524 fusion protein is shown and its wildtype predecessors. AT: AT-hook, SNL1 and SNL2: subnuclear localization sites, MT: methyltransferase, PHD1-3 and PHD4: plant homology domains, BD: binding domain, FYRC and FYRN: domains involved in heterodimerisation, TASPASE1: taspase 1 cleavage sites, TAD: transcriptional activation domain, SET: Su(var)3-9, enhancer-of-zeste trithorax domain, CC: coiled coil domain, TMD: transmembrane domain, aa: amino acids. (figure adapted from reference).
Protein
905 amino acids containing protein with transmembrane domain and coiled coil domain; inhibits MYC directed inhibitory function of protein phosphatase 2A (PP2A).

Result of the chromosomal anomaly

Hybrid gene

Description
In the described patient MLL exon 1-10 are fused to KIAA1524 exon 17-21 due to translocation between MLL intron 10 and KIAA1524 intron 16.

Transcript
See figure above.

Detection
RT-PCR according to reference.

Fusion protein

Description
From the fusion in the described patient a fusion protein of 1673 amino acids is expected.

Expression / Localisation
Unknown.

References


This article should be referenced as such: