Leukaemia Section
Short Communication

**t(11;16)(q23;q24) KMT2A/USP10**

Elena Zerkalenkova, Svetlana Lebedeva, Anna Kazakova, Pavel Baryshev, Claus Meyer, Rolf Marschalek, Galina Novichkova, Michael Maschan, Aleksey Maschan, Yulia Olshanskaya

Dmitry Rogachev Research and Clinical Center for Pediatric Hematology, Oncology and Immunology, Samora Mashela str., 1, Moscow, 117997, Russia (EZ, AK, PB, GN, MM, AM, YuO), Lomonosov Moscow State University, GSP-1, Leninskie Gory, Moscow, 119991, Russia (SL), Goethe-University of Frankfurt, Diagnostic Center of Acute Leukemia, Biocenter, Max-von-Laue-Str., 9, D-60438, Frankfurt / Main, Germany (CM, RM) / eazerkalenkova@gmail.com

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**Abstract**

Review on t(11;16)(q23;q24), with data on clinics, and the genes involved.

**Keywords**

Chromosome 11; Chromosome 16; KMT2A; USP10; Acute monoblastic leukaemia

15 years-old boy with AML-M5a relapse (Zerkalenkova et al., 2018).

**Treatment**

The patient was treated according to AML-BFM-2004 protocol and remission was achieved after the 2nd induction course. After two years a bone marrow relapse of AML M5a was diagnosed.

**Prognosis**

The prognosis was poor, the current patient died due to the progression of the disease (Zerkalenkova et al., 2018).

**Cytogenetics**

**Probes**

MLL dual color break apart rearrangement probe.

**Additional anomalies**

del(7p)
t(11;16)(q23;q24) KMT2A/USP10

Zerkalenkova E et al.

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Figure 2. The probe was hybridized to metaphase and displayed 3’-portion of KMT2A translocated to chromosome 16 (Zerkalenkova et al., 2018).

Genes involved and proteins

KMT2A (lysine (K)-specific methyltransferase 2A)

Location 11q23.3

DNA/RNA

KMT2A gene consists of 37 exons encoding a 3969 amino-acid nuclear protein with a molecular weight of nearly 431 kDa.

Protein

431 kDa; contains two DNA binding motifs (a AT hook and Zinc fingers), and a DNA methyl transferase motif; wide expression; nuclear localization; transcriptional regulatory factor.

USP10 (ubiquitin specific peptidase 10)

Location 16q24.1

DNA/RNA

USP10 gene consists of 14 exons encoding a 798 amino-acid protein with a molecular weight of 87134 Da.

Protein

87134 Da; belongs to the ubiquitin-specific proteases family; contains N-terminal thiol hydrolase catalytic domain and C-terminal Ataxin-2 extension that mediates protein-protein interactions; widely expressed; found in nucleus, cytoplasm and early endosomes; deubiquitinates various substrates. Main substrate: TP53. Upon DNA damage USP10 moves to nucleus where deubiquitinates the p53 protein thus increasing p53 levels (Yuan et al., 2010).

Result of the chromosomal anomaly

Hybrid gene

KMT2A/USP10 fusion gene was detected by LDI-PCR (Zerkalenkova et al., 2018).

KMT2A

AGCCTGGGTGACACAGTGAGACTCCATCT

KMT2A-USP10

AGCCTGGGTGACACAGCCCTGTTGTT

USP10

TGTACCCATGACCTAGCCCTGTTGTT

Figure 3.

Description

KMT2A/USP10 fusion gene contains 5’-portion of KMT2A and 3’-portion of USP10. Breakpoints are localized in KMT2A intron 12 and USP10 intron 1.

Fusion protein

Description

KMT2A/USP10 fusion protein joins amino-acid residue 1493 of KMT2A to amino-acid residue 8 of USP10 thus retaining KMT2A AT hooks, Pro-rich, and the Zn finger CXXC type domains and almost entire USP10.

References


This article should be referenced as such: