

Leukaemia Section

Short Communication

t(2;11)(p21;q23) KMT2A/?

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Abstract

Review on t(2;11)(p21;q23) with MLL (KMT2A) involvement, with data on clinics, and the genes involved.

Keywords

Chromosome 2; chromosome 11; MLL; KMT2A; acute myeloid leukemia; acute lymphoblastic leukemia

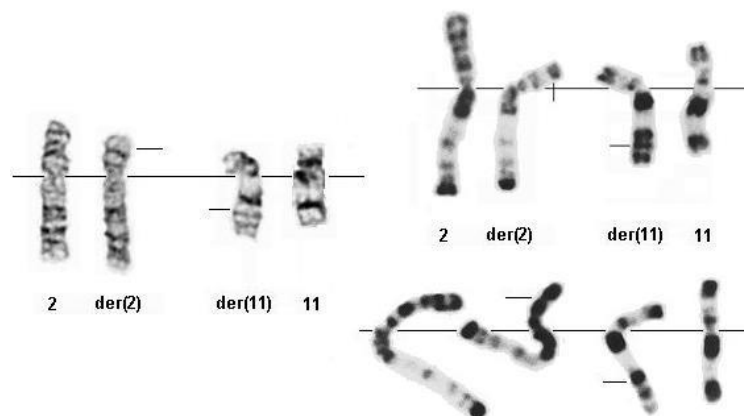
Clinics and pathology

Disease

Myelodysplastic syndromes: (MDS), acute myeloid leukemia: (AML) and acute lymphoblastic leukemia (ALL).

Phenotype/cell stem origin

Although at least 43 cases of t(2;11)(p21;q23) have been described in hematological malignancies (19 MDS, 21 AML, 2 acute lymphocytic leukemia (ALL) and 1 chronic lymphocytic leukemia: (CLL) (Mitelman et al., 2016)., The implication of MLL was ascertained in only 5 cases (Thirman et al., 1993; Finke et al., 1994; Fleischman et al., 1999; Kim et al., 2002; Meyer et al., 2006), a case with a hidden involvement of AFF3 (2q11) being discarded (Hiwatari et al., 2003), while thirty two case of t(2;11)(p21;q23) without MLL rearrangement are available (review in Ruano and Shetty, 2016). There were a case of refractory anemia with excess of blasts: (RAEB) evolving towards an AML, a M0-AML: evolving towards a M4-AML, a M5a-AML, and two ALLs.



t(2;11)(p21;q23) G- banding (left) - Courtesy Eric Crawford, and R- banding (Editor)

Epidemiology

Sex ratio was 3M/1F. Patients were aged 8 months, 58, 58, and 61 years (Thirman et al., 1993; Finke et al., 1994; Fleischman et al., 1999; Kim et al., 2002).

Clinics

variable

Cytogenetics

Additional anomalies

The t(2;11)(p21;q23) was the sole abnormality in 3 cases, and was accompanied with , del(5q) in one case. It is of note that deletions of 5q usually are not seen in cases with MLL-associated translocations.

Genes involved and proteins

Note

the gene involved in 2p in unknown

KMT2A (myeloid/lymphoid or mixed lineage leukemia)

Location

11q23.3

DNA/RNA

37 exons, spanning about 120 kb; 13-15 mRNA

Protein

3969 amino acids, 431 kDa; Transcriptional regulatory factor. MLL is known to be associated with more than 30 proteins, including the core components of the SWI/SNF chromatin remodeling complex and the transcription complex TFIID. MLL binds promoters of HOX genes through acetylation and methylation of histones. MLL is a major regulator of hematopoiesis and embryonic development, through regulation of HOX genes expression regulation (HOXA9 in particular).

Result of the chromosomal anomaly

Hybrid gene

Description

unknown

Fusion protein

Description

unknown

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