Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

t(X;11)(q22;q23)

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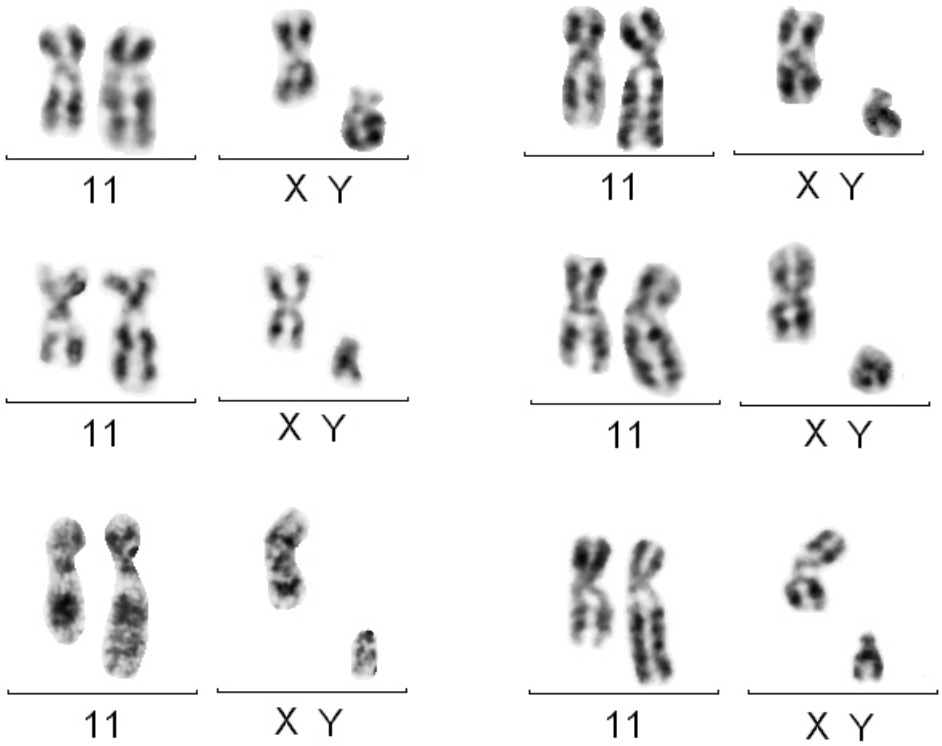

Partial karyotypes showing the chromosomal translocation t(X;11)(q22;q23).

Clinics and pathology

Disease

The chromosomal translocation t(X;11)(q22;q23) occurs very rarely, with only three cases of infants young children having been described in the literature; 2 AML cases: a 3 years old male, diagnosed with AML-M2 (Harrison et al., 1998) and 2 years old female diagnosed with acute megakaryoblastic leukemia (FAB type M7) (Ribeiro et al., 1993). The one ALL case described in a 4 years old male had a complex karyotype with chromosomal translocation t(11;14)(q13;q32), and monosomy 22 (Soszynska et al., 2008). Of note, the fourth AML (FAB type M2) case reported by Slater in a 10 months old male was shown

to involve the SEPTIN6 gene located on Xq24 (Slater et al., 2002).

Phenotype/cell stem origin

Suggested involvement of a pluripotent stem cell or a myeloid progenitor cell; myeloid lineage.

Etiology

No known prior exposure.

Epidemiology

Only 3 cases to date, sex ratio 2M/1F.

Prognosis

From the known data, the 3 years old male, diagnosed with AML-M2 remained alive in complete remission at 97 months; the ALL patient was in complete remission after 39 months.

Cytogenetics

Note

Breakpoints difficult to ascertain; cytogenetic appearance may be similar to t(X;11)(q13;q23) involving the AFX gene that fuses to MLL in acute leukemias.

Cytogenetics morphological

t(X;11)(q22;q23).

Additional anomalies

Sole abnormality in AML-M2 case, part of a hyperploid karyotype associated with +6, +8, +19, +21, +21 in a child with acute megakaryoblastic leukemia and complex karyotype in ALL case associated with t(11;14)(q13;q32), and monosomy 22, indicating that the t(X;11)(q22;q23) is likely to be a secondary anomaly to t(11;14)(q13;q32) in ALL.

Genes involved and proteins

Note

The gene in Xq22 is yet unknown, it is therefore uncertain whether this translocation involve a new MLL partner.

MLL

Location

11q23

Note

The MLL gene is frequently disrupted by a variety of chromosomal rearrangements that occur in acute myeloblastic leukemia (AML) and in acute lymphoblastic leukemia (ALL), with a peak incidence in infant leukemia as well as in secondary, topoisomerase II inhibitor-related leukemia.

DNA/RNA

The MLL genomic structure consists of 36 exons distributed over 100 kb, the mRNA of ~11.9 kb encodes a 3969 amino-acid nuclear protein with a molecular weight of of 430 kDa.

Protein

The MLL protein is a multi-domain molecule with regions of homology to diverse proteins; a major regulator of class I homeobox (HOX) gene expression.

Result of the chromosomal anomaly

Hybrid gene

Note

5' MLL - PARTNER GENE 3'.

MLL translocation breakpoints cluster within an 8.3-kb region spanning exons 5-11; genomic breakpoint junction usually created on the der(11) chromosome.

Fusion protein

Oncogenesis

Expression of a chimeric protein with actively transforming properties; altered patterns of MLL activity in hematopoietic stem cells resulting in blockage of hematopoietic maturation.

References

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