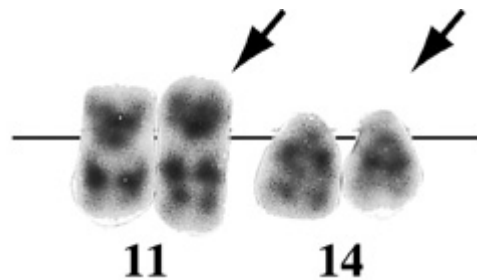


t(11;14)(q23;q24)

Identity



t(11;14)(q23;q24), G-banding

Clinics and Pathology

Disease ANLL and therapy related AL

Phenotype / cell stem origin monoblastic, unclassified

Epidemiology Rare. Three cases reported so far.

Prognosis Very poor. Less than 2 months survival in two cases.

Cytogenetics

Additional anomalies Not found in mainline in reported cases.

Genes involved and Proteins

Gene Name

[MLL](#)

Location 11q23

Dna / Rna 36 exons, spans over 100kb, ORF 12kb.

Protein 3969 amino acids; 431 kDa; contains two DNA binding motifs (a AT hook and a DNA methyltransferase homology motif), trithorax homology domains, zinc finger domains with features of PHD fingers and the C-terminal SET domain.

Gene Name

[Gephyrin \(GPHN\)](#)

Location 14q24
Dna / Rna 29 exons, spans approximately 800kb, ORF 2.3kb
Protein 736 to 770 amino acids; 93-105 kDa; submembraneous scaffold protein that anchors glycine receptor to postsynaptic cytoskeletal elements through a putative microtubule binding motif. GPHN is also involved in molybdenum cofactor biosynthesis (MoaB, MogA and MoeA homology domain), and interacts with RAFT-1.

Result of the chromosomal anomaly

Hybrid

gene 5' MLL-3' GPHN on der (11)

Description

Transcript no GPHN-MLL reciprocal transcript

Fusion Protein

MLL-GPHN protein

Description C-terminal half of GPHN, including the suspected putative microtubule binding motif and MoeA homology domain, is fused to the N-terminal portion of MLL.

External links

Other database [t\(11;14\)\(q23;q24\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(11;14\)\(q23;q24\)](#) [CancerChromosomes \(NCBI\)](#)

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:

you are welcome to submit a paper to our new [Case Report section](#).

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Contributor(s)

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