Leukaemia Section
Short Communication

\textbf{t(11;14)(q23;q24)}

Mariko Eguchi

Leukaemia Research Fund Centre, Institute of Cancer Research, Chester Beatty Laboratories, 237 Fulham Road, London SW3 6JB, UK (ME)

Published in Atlas Database: April 2002
Online updated version: \url{http://AtlasGeneticsOncology.org/Anomalies/t1114q23q24ID1198.html}
DOI: 10.4267/2042/37874

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2002 Atlas of Genetics and Cytogenetics in Oncology and Haematology

\textbf{Identity}

\textbf{Clinics and pathology}

\textbf{Disease}
ANLL and therapy related AL.

\textbf{Phenotype/cell stem origin}
Monoblastic, unclassified.

\textbf{Epidemiology}
Rare. Three cases reported so far.

\textbf{Prognosis}
Very poor. Less than 2 months survival in two cases.

\textbf{Cytogenetics}

\textbf{Additional anomalies}
Not found in mainline in reported cases.

\textbf{Genes involved and proteins}

\textbf{MLL}

\textbf{Location}
11q23

\textbf{DNA/RNA}
36 exons, spans over 100 kb, ORF 12 kb.

\textbf{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.

\textbf{Gephyrin (GPHN)}

\textbf{Location}
14q24

\textbf{DNA/RNA}
29 exons, spans approximately 800 kb, ORF 2.3 kb.

\textbf{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.

\textbf{Result of the chromosomal anomaly}

\textbf{Hybrid gene}

\textbf{Description}
5' MLL-3' GPHN on der (11).

\textbf{Transcript}
No GPHN-MLL reciprocal transcript.
**Fusion 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.

**References**


Ayton PM, Cleary ML. Molecular mechanisms of leukemogenesis mediated by MLL fusion proteins. Oncogene. 2001 Sep 10;20(40):5695-707


---

This article should be referenced as such: