

Leukaemia Section

Short Communication

t(8;11)(p11;p15)

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Published in Atlas Database: March 2005

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t0811p11p15ID1200.html>

DOI: 10.4267/2042/38193

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Clinics and pathology

Disease

Acute non lymphocytic leukemia (ANLL)

Note

Possibly heterogenous (see data on genes).

Epidemiology

3 cases to date: 1 M1-ANLL, 1 M4-ANLL and 1 case not otherwise specified; two male patients were aged 50 and 64 years.

Prognosis

One patient died during induction therapy, another one achieved complete remission and was alive at 19 months+.

Cytogenetics

Cytogenetics morphological

Sole anomaly in 1 case, accompanied with del(9q) in another case.

Genes involved and proteins

Note

In 8p11: WHSC1L1/NSD3 was proved to be implicated in the translocation in one case, while FGFR1 was (only) suspected to be involved in a second case; this case was analysed with two probes flanking FGFR1 over a distance of about 700 kb; the two probes were found to be split in FISH experiments, indicating that FGFR1 was possibly concerned by the break. However, NSD3

is 107 kb long, is at a distance of only 30 kb from FGFR1, and FGFR1 spans 56 kb; therefore, NSD3 is also a candidate in this case.

In 11p15: NUP98 was found to be implicated in the translocation in one case; in the second case, probes flanking NUP98 over a distance of about 1 Mb were used; it is likely that NUP98 is also involved in this case, although the involvement of CARS, 600 kb more telomeric than NUP98, is not excluded.

WHSC1L1/NSD3

Location

8p11

Protein

Suggested role in chromatin remodeling.

NUP98

Location

11p15

Protein

Nuclear membrane localisation; nucleoporin; associated with the nuclear pore complex; role in nucleocytoplasmic transport processes.

Result of the chromosomal anomaly

Hybrid gene

Transcript

A 5' NUP98 - 3' NSD3 fusion transcript was detected; the reciprocal transcript was also expressed. The breakpoints occurred between exons 11 and 12 of NUP98 and between exons 3 and 4 in WHSC1L1/NSD3.

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This article should be referenced as such:

Huret JL. t(8;11)(p11;p15). *Atlas Genet Cytogenet Oncol Haematol.* 2005; 9(2):164-165.
