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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 flaming 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, NDS3 is 107 kb long, is at a distance of only 30 kb from FGFR1, and FGFR1 spans 56 kb; therefore, NDS3 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 flaming 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.

References


This article should be referenced as such: