NSD1 (Nuclear receptor-binding, su(var), enhancer-of-zeste and trithorax domain-containing protein 1)

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Expression
Widely expressed.

Function
Features of a basic transcription factor, also of a bifunctional transcriptional regulator, (similar to murine Nsd1).

Homology
NSD2: (Wolf-Hirschhorn syndrome critical region on 4p); NSD3: expressed in tumour cell lines.

Implicated in
t(5;11)(q35;p15.5)/ acute non lymphoblastic leukemia (ANLL)

Disease
De novo childhood ANLL.

Prognosis
Only 5 cases reported. All had poor response to treatment/short survival.

Cytogenetics
Cryptic: associated with del(5q) (sole cytogenetic abnormality) or a normal karyotype.
5' NUP98 - 3' NSD1; NSD1-NUP98 also present in all cases tested.

Abnormal protein

NH2 NUP98- COOH NSD1: fuses the FXFG portion of NUP98 to the SET, SAC and PHD finger domains of NSD1. NSD1-NUP98: fuses the RNA-binding domain of NSD1 to the NID domain NUP98.

References


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