Gene Section
Mini Review

NUP98 (nucleoporin 98 kDa)

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**Identity**
HGNC (Hugo): NUP98
Location: 11p15

**DNA/RNA**

Transcription
3.6, 6.5, 7.0 kb mRNA.

**Protein**

Description
920 amino acids; 97 kDa; contains repeated motifs (GLFG and FG) in N-term and a RNA binding motif in C-term.

Expression
Wide.

Localisation
Nuclear membrane localisation.

Function
Nucleoporin: associated with the nuclear pore complex; role in nucleocytoplasmic transport processes.

**Homology**
Member of the GLFG nucleoporins.

**Implicated in**

inv (11)(p15q22)/ myelodysplastic syndrome (MDS) or acute non lymphocytic leukemia (ANLL) → NUP98-DDX10

Disease
Therapy related MDS (t-MDS) and ANLL; de novo ANLL.

Hybrid/Mutated gene
5'NUP98 - 3'DDX10.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the acidic domain of DDX10.

**t(1;11)(q23;p15.5)/t-MDS or ANLL → NUP98-PMX1**

Disease
One case of t-ANLL.

Hybrid/Mutated gene
5’NUP98 - 3’PMX1.
NUP98 wild type

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the homeodomain of PMX1.

\( t(2;11)(q31;p15)/ \text{treatment related leukaemia} \rightarrow \text{NUP98-HOXD13} \)

Disease
So far, only 1 case of treatment related myelodysplasia evolving towards M6 acute non lymphocytic leukaemia.

Hybrid/Mutated gene
5' NUP98 - 3' HOXD13.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the HOXD13 homeodomain.

\( t(4;11)(q21;p15.5)/ \text{T- acute lymphoblastic leukemia (ALL)} \rightarrow \text{NUP98-RAP1GDS1} \)

Disease
3 cases of adult T-ALL.

Hybrid/Mutated gene
5' NUP98 - 3' RAP1GDS1.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the entire coding region of RAP1GDS1. The product, rap1gds, has guanine nucleotide exchange factor activity.

\( t(5;11)(q35;p15.5)/ \text{ANLL} \rightarrow \text{NUP98-NSD1} \)

Disease
ANLL. 5 cases reported to date. All were children or young adults (age range 3-18 years). Note that the \( t(5;11)(q35;p15.5) \) is not detectable by G-banding. Three cases were reported as cryptic \( t(5;11) \) associated with del(5q); a further two cases were identified in apparently normal karyotypes.

Hybrid/Mutated gene
5' NUP98 - 3' NSD1.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the conserved SET, SAC and PHD finger domains of the NSD1 gene.

\( t(7;11)(p15;p15)/ \text{ANLL} \rightarrow \text{NUP98-HOXA9} \)

Disease
M2-M4 ANLL mostly; occasionally: CML-like cases.

Prognosis
Mean survival: 15 mths.

Cytogenetics
Sole anomaly most often.

Hybrid/Mutated gene
5' NUP98 - 3' HOXA9.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the HOXA9 homeobox.

\( t(9;11)(p22;p15.5)/ \text{ANLL} \rightarrow \text{NUP98-LEDGF} \)

Disease
One case of de novo ANLL.

Hybrid/Mutated gene
5' NUP98 - 3' LEDGF.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the COOH terminal of the LEDGF gene (encoding transcriptional activators p52 and p75).

\( t(11;12)(p15;q13)/ \text{treatment related leukemia (t-ANLL/MDS)} \)

Disease
1 patient with t-MDS/ANLL.

Hybrid/Mutated gene
5' NUP98 - 3' unknown.

\( t(11;17)(p15.5;q21)/ \text{t-MDS/ANLL} \)

Disease
1 patient with t-MDS/ANLL.

Hybrid/Mutated gene
5' NUP98 - 3' unknown.

\( t(11;20)(p15.5;q11)/ \text{ANLL, t-MDS/ANLL} \rightarrow \text{NUP98-TOP1} \)

Disease
ANLL, t-MDS/ANLL.

Hybrid/Mutated gene
5' NUP98 - 3' TOP1.

Abnormal protein
Fuses the GLFG repeat domains of NUP98 to the catalytic domain of TOP1.
Breakpoints

11q15 (NUP98)

1q23 (PMX1)

2q31 (HOXD13)

4q21 (RAP1GDS1)

12q13

11q22 (DDX2)

7p15 (HOXA9)

5q35 (hNSDL)

20q22 (TCP1)

Nup98 and partners. Editor 03/2002

References


This article should be referenced as such: