NUMA1 (nuclear mitotic apparatus protein 1)

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Identity

HGNC (Hugo): NUMA1
Location: 11q13

DNA/RNA

Transcription
7217 bp mRNA; coding sequence: 6305 bp.

Protein

Description
2101 amino acids; 239 kDa; the globular COOH tail domain contains a nuclear targeting sequence, a site for binding to the mitotic spindle and a site responsible for nuclear reformation; can build multiarm oligomers.

Expression
Widely expressed; also expressed in the whole embryo.

Localisation
Internal nuclear matrix protein in interphase which relocates to the spindle poles in mitosis.

Function
Component of the mitotic spindle matrix: associates with microtubule motors during mitosis; essential role in organizing microtubule minus ends at spindle poles (anchors the microtubule ends); on the other hand, may not be essential in the nucleoskeleton structural architecture during interphase.

Implicated in

\( t(11;17)(q13;q21) \)

Note
Must not be confused with the \( t(11;17)(q23;q21) \), implicating PLZF and RARA, also in M3-ANLL (see below).

Disease
Atypical M3 acute non lymphoblastic leukemia (ANLL); only 1 case fully described.

Hybrid/Mutated gene
5' exons of NuMA, fused to the exons encoding the retinoic acid and DNA-binding domains of RARA.

Abnormal protein
The NuMA-RARA fusion protein forms aggregates in the nucleus where the normal NuMA partly colocalizes.

References


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