

CPTC-NBN-1 (CAB080232)

Uniprot ID: [O60934](#)

Protein name: NBN_HUMAN

Full name: Nibrin

Tissue specificity: Ubiquitous (PubMed:9590180). Expressed at high levels in testis (PubMed:9590180).

Function: Component of the MRE11-RAD50-NBN (MRN complex) which plays a critical role in the cellular response to DNA damage and the maintenance of chromosome integrity. The complex is involved in double-strand break (DSB) repair, DNA recombination, maintenance of telomere integrity, cell cycle checkpoint control and meiosis. The complex possesses single-strand endonuclease activity and double-strand-specific 3'-5' exonuclease activity, which are provided by MRE11. RAD50 may be required to bind DNA ends and hold them in close proximity. NBN modulate the DNA damage signal sensing by recruiting PI3/PI4-kinase family members ATM, ATR, and probably DNA-PKcs to the DNA damage sites and activating their functions. It can also recruit MRE11 and RAD50 to the proximity of DSBs by an interaction with the histone H2AX. NBN also functions in telomere length maintenance by generating the 3' overhang which serves as a primer for telomerase dependent telomere elongation. NBN is a major player in the control of intra-S-phase checkpoint and there is some evidence that NBN is involved in G1 and G2 checkpoints. The roles of NBS1/MRN encompass DNA damage sensor, signal transducer, and effector, which enable cells to maintain DNA integrity and genomic stability. Forms a complex with RBBP8 to link DNA double-strand break sensing to resection. Enhances AKT1 phosphorylation possibly by association with the mTORC2 complex.

Subcellular location:

Nucleus (*experimental evidence*)

Nucleus > PML body (*experimental evidence*)

Chromosome > Telomere (*experimental evidence*)

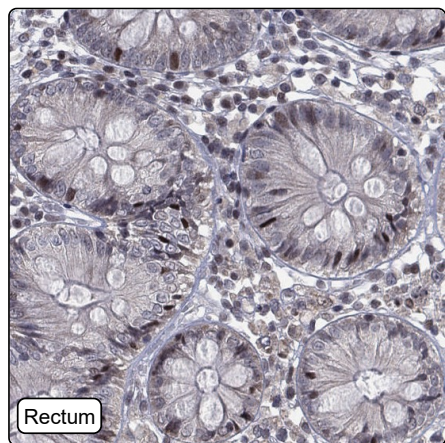
Chromosome (*experimental evidence*)

NOTE: Localizes to discrete nuclear foci after treatment with genotoxic agents (PubMed:26438602, PubMed:10783165, PubMed:26215093). Acetylation of 'Lys-5' of histone H2AX (H2AXK5ac) promotes NBN/NBS1 assembly at the sites of DNA damage (PubMed:26438602).

Protein existence: Experimental evidence at protein level

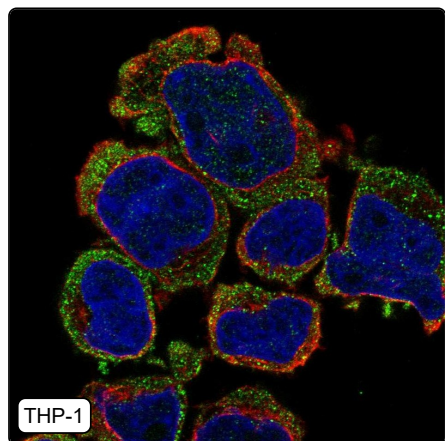
Comment:

Immunohistochemistry



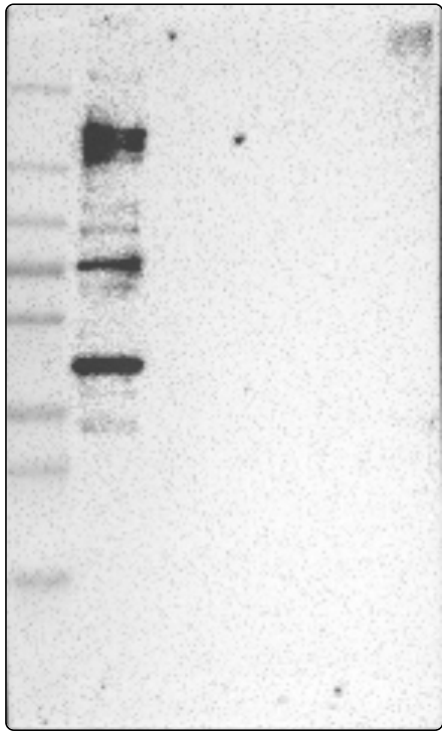
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|--------------------------|--|
| IHC protocol: | HIER pH6, Dilution 1:600 |
| IHC test staining: | Rare nuclear positivity in few tissues. Additional cytoplasmic positivity in testis. |
| Literature conformance: | Not consistent with gene/protein characterization data |
| Literature significance: | |
| RNA similarity: | Very low consistency between antibody staining and RNA expression data |
| RNA tissue specificity: | Low tissue specificity |
| RNA tissue distribution: | Detected in all |
| IHC Sibling similarity: | Other antibody shows dissimilar IHC staining pattern |
| IHC fail comment: | ANTIBODY FAILED: Improbable histological location,Not consistent with RNA |

Immunofluorescence



| | |
|-------------------------------------|--|
| IF Overlay: | antibody (green), anti-tubulin (red) and DAPI (blue) |
| IF main location: | Cytosol - 12: Uncertain (auto) |
| IF additional location: | |
| IF approved for publication on HPA: | No |
| IF in THP-1: | Cytosol |
| IF in U-2 OS: | Cytosol |

Western blot



| | |
|--------------------------------|---|
| WB Size markers (kDa): | 250, 130, 100, 70, 55, 35, 25, 15, 10 |
| WB Lanes: | Marker (1), RT4 (2), U-251 MG (3), Plasma (4), Liver (5), Tonsil (6) |
| WB Target weight (kDa): | 10, 15, 17, 18, 76, 85 |
| WB Validation: | Supported (Band of predicted size in kDa (+/-20%) with additional bands present.) |