

<b>Product name:</b>	Neurofibromin Rabbit Polyclonal Antibody
<b>Cat number:</b>	ABN14608
<b>Conjugate:</b>	Unconjugated
<b>Size:</b>	100µL
<b>Clone:</b>	Polyclonal
<b>Concentration:</b>	1mg/ml
<b>Host:</b>	Rabbit
<b>Isotype:</b>	IgG
<b>Immunogen:</b>	The antiserum was produced against synthesized peptide derived from human NF1. AA range:1551-1600
<b>Reactivity:</b>	Human,Mouse,Rat
<b>Applications:</b>	WB 1:500-1:2000,IHC 1:50-1:300
<b>Molecular Weight:</b>	319kDa
<b>Purification:</b>	Affinity purification
<b>Form:</b>	Liquid
<b>Buffer:</b>	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
<b>Storage:</b>	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

**Background:**

This gene product appears to function as a negative regulator of the ras signal transduction pathway. Mutations in this gene have been linked to neurofibromatosis type 1, juvenile myelomonocytic leukemia and Watson syndrome. The mRNA for this gene is subject to RNA editing (CGA>UGA->Arg1306Term) resulting in premature translation termination. Alternatively spliced transcript variants encoding different isoforms have also been described for this gene. [provided by RefSeq, Jul 2008], alternative products: Experimental confirmation may be lacking for some isoforms, caution: Was originally (PubMed:8807336) thought to be associated with LEOPARD (LS), an autosomal dominant syndrome., disease: Defects in NF1 are a cause of familial spinal neurofibromatosis (spinal NF) [MIM:162210]. Familial spinal NF is considered to be an alternative form of neurofibromatosis, showing multiple spinal tumors., disease: Defects in NF1 are a cause of juvenile myelomonocytic leukemia (JMML) [MIM:607785]. JMML is a pediatric myelodysplastic syndrome that constitutes approximately 30% of childhood cases of myelodysplastic syndrome (MDS) and 2% of leukemia. Germline mutations of NF1 account for the association of JMML with type 1 neurofibromatosis (NF1),., disease: Defects in NF1 are a cause of neurofibromatosis-Noonan syndrome (NFNS) [MIM:601321]. NFNS is characterized by manifestations of both NF1 and Noonan syndrome (NS). NS is a disorder characterized by dysmorphic facial features, short stature, hypertelorism, cardiac anomalies, deafness, motor delay, and a bleeding diathesis., disease: Defects in NF1 are the cause of type 1 neurofibromatosis (NF1) [MIM:162200]; also called Von Recklinghausen syndrome. NF1 is one of the most frequent autosomal dominant diseases (about 1 in 3000). It exhibits full penetrance by the age of 5 years and high mutation rate with 30 to 50% of NF1 patients representing a new mutation. Among the many clinical features of NF1 are patches of skin pigmentation (cafe-au-lait spots), Lisch nodules of the iris, peripheral nervous system associated tumors and fibromatous skin tumors., disease: Defects in NF1 are the cause of Watson syndrome (WS) [MIM:193520]. WS is characterized by the presence of pulmonary stenosis, cafe-au-lait spots, and mental retardation. WS is considered as an atypical form of NF1., disease: Defects in NF1 may be a cause of colorectal cancer (CRC) [MIM:114500]., function: Stimulates the GTPase activity of Ras. NF1 shows greater affinity for Ras GAP, but lower specific activity. May be a regulator of Ras activity., RNA editing: The stop codon (UGA) at position 1306 is created by RNA editing. Various levels of RNA editing occurs in peripheral nerve-sheath tumor samples (PNSTs) from patients with NF1. Preferentially observed in transcripts containing exon 23A., similarity: Contains 1 CRAL-TRIO domain., similarity: Contains 1 Ras-GAP domain.,