1 型神经纤维瘤抗体

产品货号： mlR4140

英文名称： NF1

中文名称： 1 型神经纤维瘤抗体

别　　名： Neurofibromin 1; DKFZp686J1293; FLJ21220; Neurofibromatosis Noonan syndrome; Neurofibromatosis related protein NF 1; Neurofibromatosis related protein NF1; neurofibromatosis type I; Neurofibromatosis-related protein NF-1; Neurofibromin 1; Neurofibromin truncated; Neurofibromin1; NF 1; NF; NF1; NF1_HUMAN; NFNS; Type 1 Neurofibromatosis; von Recklinghausen disease neurofibromin; von Recklinghausen disease related protein VRNF; VRNF; WATS; Watson disease related protein WSS; Watson syndrome; WSS.

研究领域： 肿瘤　细胞生物　免疫学　染色质和核信号　神经生物学　信号转导　表观遗传学　G 蛋白信号

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog, Horse, Rabbit,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 IF=1:100-500 （石蜡切片需做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 147/319kDa

细胞定位： 细胞核

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human Neurofibromin 1:2701-2839/2839

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

PubMed : **PubMed**

产品介绍 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].

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.

DISEASE:

Neurofibromatosis 1 (NF1) [MIM:162200]: A disease characterized by patches of skin pigmentation (cafe-au-lait spots), Lisch nodules of the iris, tumors in the peripheral nervous system and fibromatous skin tumors. Individuals with the disorder have increased susceptibility to the development of benign and malignant tumors. Note=The disease is caused by mutations affecting the gene represented in this entry.

Leukemia, juvenile myelomonocytic (JMML) [MIM:607785]: An aggressive pediatric myelodysplastic syndrome/myeloproliferative disorder characterized by malignant transformation in the hematopoietic stem cell compartment with proliferation of differentiated progeny. Patients have splenomegaly, enlarged lymph nodes, rashes, and hemorrhages. Note=The disease is caused by mutations affecting the gene represented in this entry.

Watson syndrome (WS) [MIM:193520]: A syndrome characterized by the presence of pulmonary stenosis, cafe-au-lait spots, and mental retardation. It is considered as an atypical form of neurofibromatosis. Note=The disease is caused by mutations affecting the gene represented in this entry.

Familial spinal neurofibromatosis (FSNF) [MIM:162210]: Considered to be an alternative form of neurofibromatosis, showing multiple spinal tumors. Note=The disease is caused by mutations affecting the gene represented in this entry.

Neurofibromatosis-Noonan syndrome (NFNS) [MIM:601321]: 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. Note=The disease is caused by mutations affecting the gene represented in this entry.

Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.

Similarity:

Contains 1 CRAL-TRIO domain.

Contains 1 Ras-GAP domain.

SWISS:

P21359

Gene ID:

4763

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

神经纤维素蛋白首先发现于神经细胞，是一种肿瘤抑制蛋白，通过调控 Ras 基因控制异常细胞生长，并且在 cAMP 信号传导通路中起调节作用.

神经纤维瘤Ⅰ型(neurofibromatosis type 1,NF1)是一种由内分泌紊乱引起的神经纤维瘤,属于常染色体显性遗传病，其发病率为 1/3500，主要表现为咖啡斑、神经纤维瘤、Lisch 结节(虹膜错构瘤)等。每 3，500 个新

生儿中就有一个是神经纤维细胞瘤 I 型患者，其临床表现为表皮或皮下多发性神经纤维瘤，良性多于恶性，常沿神经干分布。有时，神经纤维瘤会长大，或者发展到脑和脊髓，大约有一半以上患者智力低下。

产品图片