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Nance-Horan Syndrome Protein Rabbit pAb

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Catalog # AP57351

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q6T4R5
Reactivity	Rat, Mouse
Predicted	Pig, Dog, Human, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	179135
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Nance-Horan Syndrome Protein
Epitope Specificity	1451-1550/1651
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Nucleus.
DISEASE	Nance-Horan syndrome (NHS) [MIM:302350]: Rare X-linked disorder characterized by congenital cataracts, dental anomalies, dysmorphic features, and, in some cases, mental retardation. Distinctive dental anomalies are seen in affected males, including supernumerary incisors and crown shaped permanent teeth. Characteristic facial features are anteverted pinnae, long face, and prominent nasal bridge and nose. Carrier females display milder variable symptoms of disease with lens opacities often involving the posterior Y sutures, and on occasion dental anomalies and the characteristic facial features described. Note: The disease is caused by mutations affecting the gene represented in this entry. Cataract 40 (CTRCT40) [MIM:302200]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT40 manifests as a congenital nuclear opacity with severe visual impairment in affected males. Heterozygous females have suture cataracts and only slight reduction in vision. In some cases, cataract is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. Note: The disease is caused by mutations affecting the gene represented in this entry. Caused by copy number variations predicted to result in altered transcriptional regulation of the NHS gene: a 0.8 Mb segmental duplication-triplication encompassing the NHS, SCML1 and RAI2 genes, and an 4.8 kb intragenic deletion in NHS intron 1.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a protein containing four conserved nuclear localization signals. The encoded protein functions in eye, tooth, craniofacial and brain development, and it can regulate actin remodeling and cell morphology.

Mutations in this gene have been shown to cause Nance-Horan syndrome, and also X-linked cataract-40. Alternatively spliced transcript variants encoding different isoforms have been described for this gene. [provided by RefSeq, May 2014]

Additional Information

Gene ID	4810
Other Names	Actin remodeling regulator NHS, Congenital cataracts and dental anomalies protein, Nance-Horan syndrome protein, NHS
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	NHS
Function	May function in cell morphology by maintaining the integrity of the circumferential actin ring and controlling lamellipod formation. Involved in the regulation eye, tooth, brain and craniofacial development.
Cellular Location	[Isoform 1]: Apical cell membrane; Peripheral membrane protein. Cell projection, lamellipodium. Cell junction, tight junction. Cell junction, focal adhesion. Note=Colocalizes with the tight junction protein TJP1 in epithelial cells. Localizes to the leading edge of lamellipodia in motile cells
Tissue Location	Detected at low levels in all tissues analyzed. Detected in fetal and adult brain, lens, retina, retinal pigment epithelium, placenta, lymphocytes and fibroblasts. Levels in retinal pigment epithelium, placenta, lymphocytes, and fibroblasts are very low. Expressed also in kidney, lung and thymus

Background

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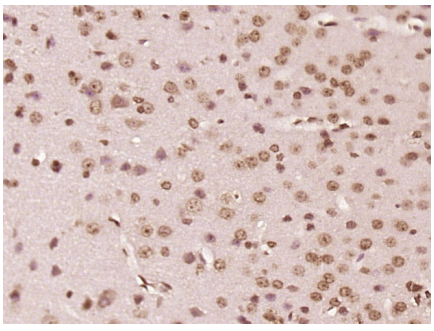
Images



Paraformaldehyde-fixed, paraffin embedded (Mouse brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (Nance-Horan Syndrome Protein) Polyclonal Antibody, Unconjugated (AP57351) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.