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PAX3 Rabbit pAb

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Catalog # AP52171

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	P23760
Reactivity	Human, Mouse, Rat
Host	Rabbit
Clonality	Polyclonal
Calculated MW	52968
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PAX3
Epitope Specificity	151-250/479
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.
SIMILARITY	Belongs to the paired homeobox family. Contains 1 homeobox DNA-binding domain. Contains 1 paired domain.
SUBUNIT	Can bind to DNA as a homodimer or a heterodimer with PAX7. Interacts with PAXBP1; the interaction links PAX3 to a WDR5-containing histone methyltransferase complex. Interacts with DAXX.
DISEASE	Waardenburg syndrome 1 (WS1) [MIM:193500]: WS1 is an autosomal dominant disorder characterized by non-progressive sensorineural deafness, pigmentary disturbances such as frontal white blaze of hair, heterochromia of irides, white eyelashes, leukoderma, and wide bridge of nose owing to lateral displacement of the inner canthus of each eye (dystopia canthorum). WS1 shows variable clinical expression and some affected individuals do not manifest hearing impairment or iris pigmentation disturbances. Dystopia canthorum is the most consistent sign and is found in 98% of the patients. Note=The disease is caused by mutations affecting the gene represented in this entry. Waardenburg syndrome 3 (WS3) [MIM:148820]: WS3 is an autosomal dominant disorder characterized by sensorineural deafness, pigmentary disturbances, dystopia canthorum and limb anomalies such as hypoplasia of the musculoskeletal system, flexion contractures, fusion of the carpal bones, syndactylies. Note=The disease is caused by mutations affecting the gene represented in this entry. Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. Note=The disease is caused by mutations affecting the gene represented in this entry. Rhabdomyosarcoma 2 (RMS2) [MIM:268220]: A form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines. Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in

	children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration involving PAX3 is found in rhabdomyosarcoma. Translocation (2;13)(q35;q14) with FOXO1. The resulting protein is a transcriptional activator. Note=A chromosomal aberration involving PAX3 is a cause of rhabdomyosarcoma. Translocation t(2;2)(q35;p23) with NCOA1 generates the NCOA1-PAX3 oncogene consisting of the N-terminus part of PAX3 and the C-terminus part of NCOA1. The fusion protein acts as a transcriptional activator. Rhabdomyosarcoma is the most common soft tissue carcinoma in childhood, representing 5-8% of all malignancies in children.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This protein is a member of the paired box (PAX) family of transcription factors. Members of the PAX family typically contain a paired box domain and a paired-type homeodomain. These proteins play critical roles during fetal development. Mutations in paired box gene 3 are associated with Waardenburg syndrome, craniofacial-deafness-hand syndrome, and alveolar rhabdomyosarcoma. The translocation t(2;13)(q35;q14), which represents a fusion between PAX3 and the forkhead gene, is a frequent finding in alveolar rhabdomyosarcoma. Alternative splicing results in transcripts encoding isoforms with different C-termini.

Additional Information

Gene ID	5077
Other Names	Paired box protein Pax-3, HuP2, PAX3, HUP2
Dilution	WB=1:500-2000,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	PAX3
Synonyms	HUP2
Function	Transcription factor that may regulate cell proliferation, migration and apoptosis. Involved in neural development and myogenesis. Transcriptional activator of MITF, acting synergistically with SOX10 (PubMed:21965087).
Cellular Location	Nucleus.

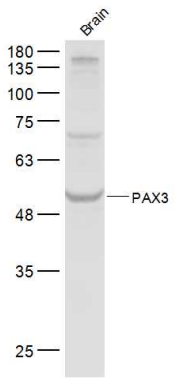
Background

This protein is a member of the paired box (PAX) family of transcription factors. Members of the PAX family typically contain a paired box domain and a paired-type homeodomain. These proteins play critical roles during fetal development. Mutations in paired box gene 3 are associated with Waardenburg syndrome, craniofacial-deafness-hand syndrome, and alveolar rhabdomyosarcoma. The translocation t(2;13)(q35;q14), which represents a fusion between PAX3 and the forkhead gene, is a frequent finding in alveolar rhabdomyosarcoma. Alternative splicing results in transcripts encoding isoforms with different C-termini.

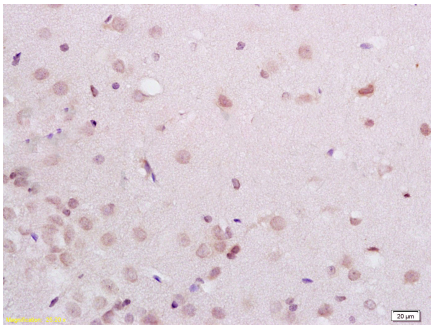
References

Goulding M.D.,et al.EMBO J. 10:1135-1147(1991).
Carninci P.,et al.Science 309:1559-1563(2005).
Epstein D.J.,et al.Cell 67:767-774(1991).
Diao Y.,et al.Cell Stem Cell 11:231-241(2012).
Vogan K.J.,et al.Genomics 17:364-369(1993).

Images



Sample:
Brain (Mouse) Lysate at 40 ug
Primary: Anti-PAX3 (AP52171) at 1/300 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 53 kD
Observed band size: 53 kD



Tissue/cell: rat brain tissue; 4% Paraformaldehyde-fixed and paraffin-embedded;
Antigen retrieval: citrate buffer (0.01M, pH 6.0), Boiling bathing for 15min; Block endogenous peroxidase by 3% Hydrogen peroxide for 30min; Blocking buffer (normal goat serum,C-0005) at 37℃ for 20 min;
Incubation: Anti-PAX3 Polyclonal Antibody, Unconjugated(AP52171) 1:200, overnight at 4°C, followed by conjugation to the secondary antibody(SP-0023) and DAB(C-0010) staining