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NR0B1 Rabbit pAb

Catalog # AP99079

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	P51843
Other Accession	P51843
Predicted	Rat, Pig, Human, Mouse, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	51718
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human NR0B1
Epitope Specificity	331-430/470
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the nuclear hormone receptor family. NR0 subfamily.
SUBUNIT	Homodimer. Interacts with NR5A1, NR5A2, NR0B2 and with COPS2.
DISEASE	Defects in NR0B1 are the cause of X-linked adrenal hypoplasia congenital (XL-AHC) [MIM:300200]; also known as X-linked Addison disease (AHX). XL-AHC is a developmental disorder of the adrenal gland that results in profound hormonal deficiencies and is lethal if untreated. It is characterized by the absence of the permanent zone of the adrenal cortex and by a structural disorganization of the glands. Hypogonadotropic hypogonadism (HHG) is frequently associated with this disorder. HHG is a condition resulting from or characterized by abnormally decreased gonadal function, with retardation of growth and sexual development. Defects in NR0B1 are the cause of 46,XY sex reversal type 2 (SRXY2) [MIM:300018]. It is a condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Note=XY individuals with a duplication of part of the short arm of the X chromosome and an intact SRY gene develop as females. The single X chromosome in these individuals does not undergo X-chromosome inactivation; therefore, these individuals presumably carry 2 active copies of genes, including the NR0B1 gene, in the duplicated region. Individuals with deletion of this region develop as males. Genes within the dosage-sensitive sex reversal region are, therefore, not essential for testis development, but, when present in a double dose, interfere with testis formation.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Adrenal hypoplasia congentia (AHC) is an X-linked disorder characterized by primary adrenal insufficiency. The disorder, which is lethal if untreated, results in adrenal insufficiency early in infancy and is characterized by low serum concentration of glucocorticoids, mineralcorticoids and androgens and failure to respond to ACTH. AHC has been mapped to chromosome Xp21 at the same or close to an X-linked locus involved in sex determination, DSS (for dosage-sensitive sex reversal). The gene corresponding to DSS and AHC (designated DAX-1 for DSS-AHC critical region on the X chromosome, gene 1) has been cloned and shown to be deleted in AHC deletion patients and

mutated in AHC non-deletion patients. The carboxy terminal 250 amino acids of the DAX-1-encoded protein, DAX-1, exhibits approximately 50% continuous similarity to the ligand-binding domain of the members of the nuclear hormone receptor superfamily while the amino terminal domain contains a putative DNA-binding motif. DAX-1 binds to retinoic acid responsive elements and down regulates retinoic acid receptor-mediated transcriptional activation.

Additional Information

Gene ID	190
Other Names	CK820_G0033312; DAX1; AHC; AHCH; AHX; DAX-1; DSS; GTD; HHG; NROB1; SRXY2; mDax1-134; mDax1-218; mDax1-225; mDax1-249; mDax1-292; mDax1-297; mDax1-300; mDax1-308; mDax1-318; mDax1-329; mDax1-337; mDax1-344; mDax1-38; mDax1-404; mDax1-472; NR0B1_PANTR; NR0B1; Nuclear receptor DAX-1; NR0B1_HUMAN; DSS-AHC critical region on the X chromosome protein 1; NR0B1_MOUSE; nuclear receptor subfamily 0 group B member 1; dosage-sensitive sex reversal; nuclear receptor subfamily 0, group B, member 1
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	NR0B1
Synonyms	AHC, DAX1 {ECO:0000303|PubMed:26416531}
Function	Nuclear receptor that lacks a DNA-binding domain and acts as a corepressor that inhibits the transcriptional activity of other nuclear receptors through heterodimeric interactions (PubMed:12482977, PubMed:32433991). Component of a cascade required for the development of the hypothalamic-pituitary-adrenal-gonadal axis (PubMed:7990953, PubMed:8675564). May also have a role in the development of the embryo and in the maintenance of embryonic stem cell pluripotency (By similarity).
Cellular Location	Nucleus. Cytoplasm. Note=Shuttles between the cytoplasm and nucleus. Homodimers exits in the cytoplasm and in the nucleus

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