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KMT3B Rabbit pAb

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Catalog # AP58904

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q96L73
Reactivity	Rat, Human, Mouse
Predicted	Pig, Dog, Rabbit, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	296652
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human KMT3B/NSD1/ARA267
Epitope Specificity	2401-2600/2696
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. Chromosome (Probable).
DISEASE	Defects in NSD1 are the cause of Sotos syndrome type 1(SOTOS1) [MIM:117550]; also known as cerebral gigantism. It is adisorder characterized by excessively rapid growth, acromegalicfeatures, and a nonprogressive cerebral disorder with mentalretardation. High-arched palate and prominent jaw are noted inseveral patients. Most cases of Sotos syndrome are sporadic and mayrepresent new dominant mutation.Defects in NSD1 are the cause of Weaver syndrome type 1(WVS1) [MIM:277590]. A syndrome of accelerated growth and osseousmaturation, unusual craniofacial appearance, hoarse and low-pitchedcry, and hypertonia with camptodactyly. Distinguishing features ofWeaver syndrome include broad forehead and face, ocularhypertelorism, prominent wide philtrum, micrognathia, deephorizontal chin groove, and deep-set nails. In addition, carpalbone development is advanced over the rest of the hand.Defects in NSD1 are a cause of Beckwith-Wiedemannsyndrome (BWS) [MIM:130650]. BWS is a genetically heterogeneousdisorder characterized by anterior abdominal wall defects includingexomphalos (omphalocele), pre- and postnatal overgrowth, andmacroglossia. Additional less frequent complications includespecific developmental defects and a predisposition to embryonaltumors.Note=A chromosomal aberration involving NSD1 is found inchildhood acute myeloid leukemia. Translocation t(5;11)(q35;p15.5)with NUP98.Note=A chromosomal aberration involving NSD1 is found inan adult form of myelodysplastic syndrome (MDS). Insertion of NUP98into NSD1 generates a NUP98-NSD1 fusion product.
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 a SET domain, 2 LXXLL motifs, 3 nuclear translocation signals (NLSs), 4 plant homeodomain (PHD) finger regions, and a proline-rich region. The encoded protein enhances androgen receptor (AR) transactivation, and this enhancement can be increased further in the presence of other androgen receptor associated coregulators. This

protein may act as a nucleus-localized, basic transcriptional factor and also as a bifunctional transcriptional regulator. Mutations of this gene have been associated with Sotos syndrome and Weaver syndrome. One version of childhood acute myeloid leukemia is the result of a cryptic translocation with the breakpoints occurring within nuclear receptor-binding Su-var, enhancer of zeste, and trithorax domain protein 1 on chromosome 5 and nucleoporin, 98-kd on chromosome 11. Two transcript variants encoding distinct isoforms have been identified for this gene. [provided by RefSeq, Jul 2008].

Additional Information

Gene ID	64324
Other Names	Histone-lysine N-methyltransferase, H3 lysine-36 specific, 2.1.1.357, Androgen receptor coactivator 267 kDa protein, Androgen receptor-associated protein of 267 kDa, H3-K36-HMTase, Lysine N-methyltransferase 3B, Nuclear receptor-binding SET domain-containing protein 1, NR-binding SET domain-containing protein, NSD1, ARA267, KMT3B
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	NSD1
Synonyms	ARA267, KMT3B
Function	Histone methyltransferase that dimethylates Lys-36 of histone H3 (H3K36me2). Transcriptional intermediary factor capable of both negatively or positively influencing transcription, depending on the cellular context.
Cellular Location	Nucleus. Chromosome.
Tissue Location	Expressed in the fetal/adult brain, kidney, skeletal muscle, spleen, and the thymus, and faintly in the lung

Background

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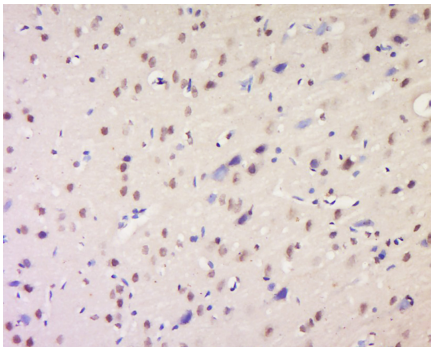
Images

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-KMT3B Polyclonal Antibody, Unconjugated(AP58904) 1:200, overnight at 4°C, followed by conjugation to the secondary antibody(SP-0023) and DAB(C-0010) staining