

Product datasheet

Recombinant human KMT3B / NSD1 protein ab196419

2 Images

Description

Product name	Recombinant human KMT3B / NSD1 protein
Purity	>= 76 % SDS-PAGE.
Expression system	Escherichia coli
Accession	<u>Q96L73</u>
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	KAQKELRQLQED RKNDKKPPPY KHIKVNRP RVQIFTADLS EIPRCNCKAT DENPCGIDSE CINRMLLYEC HPTVCPAGGR CQNQCFSKRQ YPEVEIFRTL QRGWGLRKT DIKKGEFVNE YGELIDEE CRARIRYAQE HDITNFYMLT LDKDRIIDAG PKGNARFMN HCCQPNCEQ KWSVNGDTRV GLFALSDIKA GTELTFNYNL ECLGNGKTVC KCGAPNCSGF LGVPRKNQPIATEE
Predicted molecular weight	54 kDa including tags
Amino acids	1849 to 2094
Tags	GST tag N-Terminus
Additional sequence information	NM_022455.

Specifications

Our **Abpromise guarantee** covers the use of **ab196419** in the following tested applications.

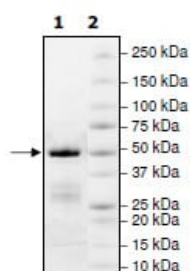
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Functional Studies SDS-PAGE
Form	Liquid

Preparation and Storage

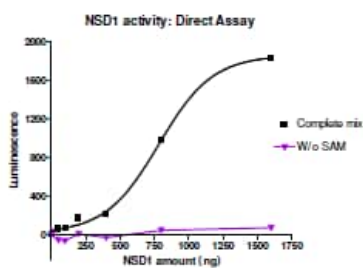
Stability and Storage	<p>Shipped on Dry Ice. Store at -80°C. Avoid freeze / thaw cycle.</p> <p>pH: 8.00</p> <p>Constituents: 0.63% Tris HCl, 0.64% Sodium chloride, 0.02% Potassium chloride, 0.12% Glutathione, 20% Glycerol (glycerin, glycerine), 0.05% (R*,R*)-1,4-Dimercaptobutan-2,3-diol</p> <p>This product is an active protein and may elicit a biological response in vivo, handle with caution.</p>
General Info	
Function	Histone methyltransferase. Preferentially methylates 'Lys-36' of histone H3 and 'Lys-20' of histone H4 (in vitro). Transcriptional intermediary factor capable of both negatively or positively influencing transcription, depending on the cellular context.
Tissue specificity	Expressed in the fetal/adult brain, kidney, skeletal muscle, spleen, and the thymus, and faintly in the lung.
Involvement in disease	<p>Defects in NSD1 are the cause of Sotos syndrome (SOTOSS) [MIM:117550]; also known as cerebral gigantism. It is a disorder characterized by excessively rapid growth, acromegalic features, and a nonprogressive cerebral disorder with mental retardation. High-arched palate and prominent jaw are noted in several patients. Most cases of Sotos syndrome are sporadic and may represent new dominant mutation.</p> <p>Defects in NSD1 are the cause of Weaver syndrome (WES) [MIM:277590]. WES is a syndrome of accelerated growth and osseous maturation, unusual craniofacial appearance, hoarse and low-pitched cry, and hypertonia with camptodactyly.</p> <p>Defects in NSD1 are a cause of Beckwith-Wiedemann syndrome (BWS) [MIM:130650]. BWS is a genetically heterogeneous disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors.</p> <p>Note=A chromosomal aberration involving NSD1 is found in childhood acute myeloid leukemia. Translocation t(5;11)(q35;p15.5) with NUP98.</p> <p>Note=A chromosomal aberration involving NSD1 is found in an adult form of myelodysplastic syndrome (MDS). Insertion of NUP98 into NSD1 generates a NUP98-NSD1 fusion product.</p>
Sequence similarities	<p>Belongs to the histone-lysine methyltransferase family.</p> <p>Contains 1 AWS domain.</p> <p>Contains 4 PHD-type zinc fingers.</p> <p>Contains 1 post-SET domain.</p> <p>Contains 2 PWWP domains.</p> <p>Contains 1 SET domain.</p>
Cellular localization	Nucleus. Chromosome.

Images



SDS-PAGE - Recombinant human KMT3B / NSD1 protein (ab196419)

SDS-PAGE of 3µg ab196419 (lane 1). Protein marker (lane 2).
4-20% gradient gel, coomassie staining.



Functional Studies - Recombinant human KMT3B / NSD1 protein (ab196419)

Activity assay of ab196419. Assay was performed using a NSD1 Chemiluminescent Assay Kit with 400 µM S-adenosyl- methionine on biotinylated H3 peptide coated plates overnight at room temperature.

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