

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	Q96L73
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	<p>KAQKELRQLQED RKNDKKPPPY KHIKVNRP RVQIFTADLS EIPRCNCKAT DENPCGIDSE CINRM LLYEC HPTVCPAGGR CQNQCFSKRQ YPEVEIFR TL QRGWGLRKT DIKKGEFVNE YGELIDEE CRARIRYAQE HDITNFYMLT LDKDRIIDAG PKG NYARFMN HCCQPCETQ KWSVNGDTRV GLFALS DIKA GTELTFNYNL ECLGNGKTVC KCGAPNC SGF LGVRPKNQPIATEE</p>
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.

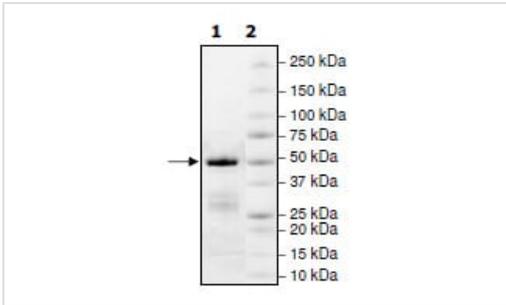
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Functional Studies SDS-PAGE
Form	Liquid

Preparation and Storage

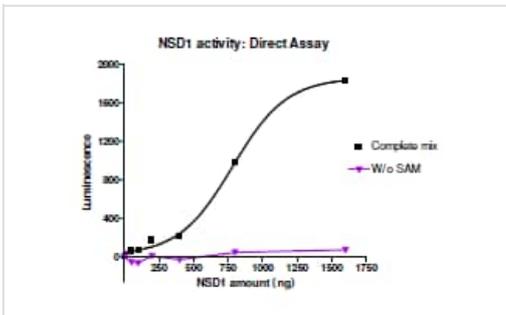
Stability and Storage	<p>Shipped on Dry Ice. Store at -80°C. Avoid freeze / thaw cycle.</p> <p>pH: 8.00</p> <p>Constituents: 0.63% Tris HCl, 0.64% Sodium chloride, 0.02% Potassium chloride, 0.12% Glutathione, 20% Glycerol, 0.05% DTT</p> <p>This product is an active protein and may elicit a biological 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 of 3µg ab196419 (lane 1). Protein marker (lane 2).
4-20% gradient gel, coomassie staining.

SDS-PAGE - 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.

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

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