

Rabbit Anti-EFTUD2 antibody

SL13061R

Product Name:	EFTUD2
Chinese Name:	延伸因子Binding proteinEFTUD2抗体
Alias:	116 kDa; 116 kDa U5 small nuclear ribonucleoprotein component; EFTUD2; Elongation factor Tu GTP binding domain containing 2; Elongation factor Tu GTP-binding domain-containing protein 2; hSNU114; MFDM; Snrp116; Snu114; SNU114 homolog; U5 116KD; U5 small nuclear ribonucleoprotein component; U5 snRNP specific protein, 116 kD; U5 snRNP specific protein, 116 kDa; U5 snRNP-specific protein; U5-116 kDa; U5S1 HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Chicken, Dog, Pig, Cow, Horse, Rabbit, Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	109kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EFTUD2:901-972/972
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20 °C. 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.
PubMed:	<u>PubMed</u>
Product Detail:	Spliceosomes are multi-protein complexes that are composed of snRNPs (small nuclear ribonucleoproteins) and a variety of associated protein factors, all of which work in

concert to regulate the splicing of pre-mRNA. Snrp116, also known as EFTUD2 (elongation factor Tu GTP binding domain containing 2) or Snu114, is a 972 amino acid protein that localizes to the nucleus and belongs to the GTP-binding elongation factor family. Existing as a component of the multi-protein U5 snRNP spliceosome complex, Snrp116 plays an important role in pre-mRNA splicing, as well as in the recycling of spliceosomal snRNPs. The gene encoding Snrp116 maps to human chromosome 17, which comprises over 2.5% of the human genome and encodes over 1,200 genes.

Function:

Component of the U5 snRNP complex required for pre-mRNA splicing. Binds GTP.

Subunit:

Identified in the spliceosome C complex. Interacts with ERBB4 and PRPF8.

Subcellular Location:

Nucleus.

Post-translational modifications:

Phosphorylated upon DNA damage, probably by ATM or ATR.

DISEASE:

Defects in EFTUD2 are the cause of mandibulofacial dysostosis with microcephaly (MFDM) [MIM:610536]. A rare syndrome characterized by progressive microcephaly, midface and malar hypoplasia, micrognathia, microtia, dysplastic ears, preauricular skin tags, significant developmental delay, and speech delay. Many patients have major sequelae, including choanal atresia that results in respiratory difficulties, conductive hearing loss, and cleft palate.

Similarity:

Belongs to the GTP-binding elongation factor family. EF-G/EF-2 subfamily.

SWISS:

O15029

Gene ID:

9343

Database links:

Entrez Gene: 428281Chicken

Entrez Gene: 509425Cow

Entrez Gene: 9343Human

Entrez Gene: 20624Mouse

Entrez Gene: 287739Rat

Entrez Gene: 379223 Xenopus laevis

Entrez Gene: 594937Xenopus tropicalis

Entrez Gene: 393480Zebrafish

Omim: 603892Human

SwissProt: Q5F3X4Chicken

SwissProt: A4FUD3Cow

SwissProt: Q15029Human

SwissProt: O08810Mouse

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.