abcam

Product datasheet

Recombinant Human SIL1 protein ab128448

1 Image

Description

Product name	Recombinant Human SIL1 protein
Purity	> 90 % SDS-PAGE. ab128448 was purified using conventional chromatography.
Expression system	Escherichia coli
Accession	Q9H173
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MHQNLKEFAL TNPEKSSTKE TERKETKAEE ELDAEVLEVF HPTHEWQALQ PGQAVPAGSH VRLNLQTGER EAKLQYEDKF RNNLKGKRLD INTNTYTSQD LKSALAKFKE GAEMESSKED KARQAEVKRL FRPIEELKKD FDELNVVIET DMQIMVRLIN KFNSSSSSLE EKIAALFDLE YYVHQMDNAQ DLLSFGGLQV VINGLNSTEP LVKEYAAFVL GAAFSSNPKV QVEAIEGGAL QKLLVILATE QPLTAKKKVL FALCSLLRHF PYAQRQFLKL GGLQVLRTLV QEKGTEVLAV RVVTLLYDLV TEKMFAEEEA ELTQEMSPEK LQQYRQVHLL PGLWEQGWCE ITAHLLALPE HDAREKVLQT LGVLLTTCRD RYRQDPQLGR TLASLQAEYQ VLASLELQDG EDEGYFQELL GSVNSLLKEL RLEHHHHHH
Predicted molecular weight	50 kDa
Amino acids	32 to 461
Tags	His tag C-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab128448** in the following tested applications.

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

Applications	SDS-PAGE

Form	Liquid

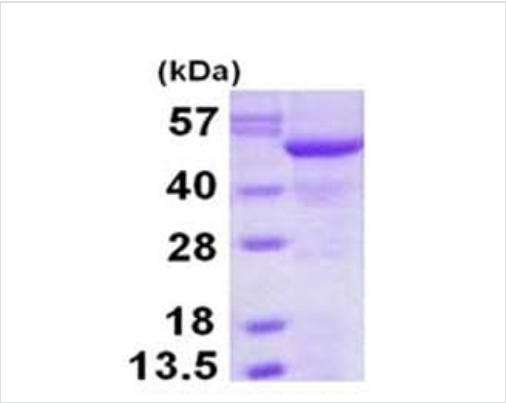
Preparation and Storage

Stability and Storage	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -80°C. Avoid freeze / thaw cycle. pH: 8.00 Constituents: 0.32% Tris HCl, 10% Glycerol (glycerin, glycerine), 0.58% Sodium chloride

General Info

Function	Required for protein translocation and folding in the endoplasmic reticulum (ER). Functions as a nucleotide exchange factor for the ER lumenal chaperone HSPA5.
Tissue specificity	Highly expressed in tissues which produce large amounts of secreted proteins such as kidney, liver and placenta. Also expressed in colon, heart, lung, ovary, pancreas, peripheral leukocyte, prostate, spleen and thymus. Expressed at low levels throughout the brain.
Involvement in disease	Defects in SIL1 are a cause of Marinesco-Sjoegren syndrome (MSS) [MIM:248800]. MSS is an autosomal recessive multisystem disorder which is characterized by cerebellar ataxia due to cerebellar atrophy, with Purkinje and granule cell loss and myopathy featuring marked muscle replacement with fat and connective tissue. Other cardinal features include bilateral cataracts, hypergonadotrophic hypogonadism and mild to severe mental retardation. Skeletal abnormalities, short stature, dysarthria, strabismus and nystagmus are also frequent findings. Mutational inactivation of this protein may result in ER stress-induced cell death signaling or malfunctioning chaperone machineries that mishandle client proteins which are critical for the organs targeted in MSS.
Sequence similarities	Belongs to the SIL1 family.
Developmental stage	Expressed in fetal kidney, fetal lung, fetal liver and at low levels in fetal brain.
Post-translational modifications	N-glycosylated.
Cellular localization	Endoplasmic reticulum lumen.

Images



SDS-PAGE - Recombinant Human SIL1 protein (ab128448)

15% SDS-PAGE analysis of ab128448 (3 µg).

Our Abpromise to you: Quality guaranteed and expert technical support

- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit **https://www.abcam.com/abpromise** or contact our technical team.

Terms and conditions

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors