Recombinant Human SLC19A3 Protein, MYC/DDK-tagged, C13 and N15-labeled

Cat. No. SLC19A3-177H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	SLC19A3 MS Standard C13 and N15-labeled recombinant protein (NP_079519) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
Species	Human
Source	HEK293
Description	This gene encodes a ubiquitously expressed transmembrane thiamine transporter that lacks folate transport activity. Mutations in this gene cause biotin-responsive basal ganglia disease (BBGD); a recessive disorder manifested in childhood that progresses to chronic encephalopathy, dystonia, quadriparesis, and death if untreated. Patients with BBGD have bilateral necrosis in the head of the caudate nucleus and in the putamen. Administration of high doses of biotin in the early progression of the disorder eliminates pathological symptoms while delayed treatment results in residual paraparesis, mild cognitive disability, or dystonia. Administration of thiamine is ineffective in the treatment of this disorder. Experiments have failed to show that this protein can transport biotin. Mutations in this gene also cause a Wernicke's-like encephalopathy. [provided by RefSeq, Jan 2010]
Molecular Mass	55.7 kDa
AA Sequence	MDCYRTSLSSSWIYPTVILCLFGFFSMMRPSEPFLIPYLSGPDKNLTSAEITNEIFPVW TYSYLVLLLPVFVLTDYVRYKPVIILQGISFIITWLLLLFGQGVKTMQVVEFFYGMVTAA

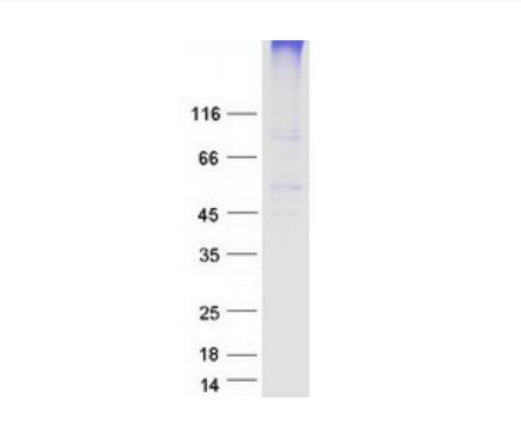
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	EVAYYAYIYSVVSPEHYQRVSGYCRSVTLAAYTAGSVLAQLLVSLANMSYFYLNVISL ASVSVAFLFSLFLPMPKKSMFFHAKPSREIKKSSSVNPVLEETHEGEAPGCEEQKPT SEILSTSGKLNKGQLNSLKPSNVTVDVFVQWFQDLKECYSSKRLFYWSLWWAFATA GFNQVLNYVQILWDYKAPSQDSSIYNGAVEAIATFGGAVAAFAVGYVKVNWDLLGEL ALVVFSVVNAGSLFLMHYTANIWACYAGYLIFKSSYMLLITIAVFQIAVNLNVERYALV FGINTFIALVIQTIMTVIVVDQRGLNLPVSIQFLVYGSYFAVIAGIFLMRSMYITYSTKSQ KDVQSPAPSENPDVSHPEEESNIIMSTKLTRTRPLEQKLISEEDLAANDILDYKDDDD KV	
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining
Stability	Stable for 3 months from receipt of products under proper storage and handling conditions.
Storage	Store at -80 centigrade. Avoid repeated freeze-thaw cycles.
Concentration	50 μg/mL as determined by BCA
Storage Buffer	100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name	SLC19A3 solute carrier family 19 member 3 [Homo sapiens (human)]
Official Symbol	SLC19A3
Synonyms	SLC19A3; solute carrier family 19 member 3; BBGD; THMD2; THTR2; thiamine transporter 2; solute carrier family 19 (thiamine transporter), member 3
Gene ID	80704

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mRNA Refseq	NM_025243
Protein Refseq	NP_079519
MIM	606152
UniProt ID	Q9BZV2
SDS-PAGE	

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