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Product Information

MemDX™ Membrane Protein Human SLC19A3 (Solute carrier family 19 member 3) Full Length

Cat. No.: **MPC0759K**
This product is for research use only and is not intended for diagnostic use.

This product is a 55.6 kDa Human SLC19A3 membrane protein expressed in HEK293. The protein is for research use only and is not approved for use in humans or in clinical diagnosis.

Product Specifications

Host Species
Human

Target Protein
SLC19A3

Protein Length
Full length

Protein Class
Transporter

Molecular Weight
55.6 kDa

TMD
12

Sequence
MDCYRTSLSSSWIYPTVILCLFGFFSMMRPSEPFLIPYLSGPDKNLTSAE
ITNEIFPVWTYSYLVLLLPVFVLTDYVRYKPVIILQGISFIITWLLLLFG
QGVKTMQVVEFFYGMVTAAEVAYYAYIYSVVSPEHYQRVSGYCRSVTLAA
YTAGSVLAQLLVSLANMSYFYLNVISLASVSVAFLFSLFLPMPKKSMFFH
AKPSREIKKSSSVNPVLEETHEGEAPGCEEQKPTSEILSTSGKLNKGQLN
SLKPSNVTVDVFVQWFQDLKECYSSKRLFYWSLWWAFATAGFNQVLNYVQ
ILWDYKAPSQDSSIYNGAVEAIATFGGAVAAFAVGYVKVNWDLLGELALV
VFSVVNAGSLFLMHYTANIWACYAGYLIFKSSYMLLITIAVFQIAVNLNV
ERYALVFGINTFIALVIQTIMTVIVVDQRGLNLPVSIQFLVYGSYFAVIA
GIFLMRSMYITYSTKSQKDVQSPAPSENPDVSHPEEESNIIMSTKL

Product Description

Expression Systems
HEK293

Tag

Based on specific requirements

Protein Format

Detergent or based on specific requirements

Form

Liquid

Storage

Aliquot and store at -20°C or lower. For long term storage, we recommend to store at -72°C or lower. Avoid freeze/thaw cycles.

Target

Target Protein

SLC19A3

Full Name

Solute carrier family 19 member 3

Introduction

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.

Alternative Names

BBGD; THMD2; THTR2; thTr-2; thiamine transporter 2; solute carrier family 19 (thiamine transporter), member 3; SLC19A3; Solute carrier family 19 member 3

Gene ID

80704

UniProt ID

Q9BZV2