www.bioss.com.cn
sales@bioss.com.cn
techsupport@bioss.com.cn
400-901-9800

SLC19A3 Antibody Blocking Peptide

Catalog Number: bs-8702P
Activity: Not tested
Purification: HPLC
Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.
Background: 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 mental retardation, 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]