Human FUCA1 Protein (His Tag)

Catalog Number: **13893-H08H**

General Information

Gene Name Synonym:

FUCA

Protein Construction:

A DNA sequence encoding the human FUCA1 (P04066) (Gln32-Lys466) was expressed with a polyhistidine tag at the C-terminus.

Source: Human

Expression Host: HEK293 Cells

QC Testing

Purity: > 95 % as determined by SDS-PAGE

Endotoxin:

< 1.0 EU per μg of the protein as determined by the LAL method

Stability:

Samples are stable for up to twelve months from date of receipt at -70 ℃

Predicted N terminal: Gln 32

Molecular Mass:

The recombinant human FUCA1 consists of 446 amino acids and predicts a molecular mass of 51.9 KDa. It migrates as an approximately 57 KDa band in SDS-PAGE under reducing conditions.

Formulation:

Lyophilized from sterile PBS, pH 7.4

Normally 5 % - 8 % trehalose, mannitol and 0.01% Tween80 are added as protectants before lyophilization. Specific concentrations are included in the hardcopy of COA. Please contact us for any concerns or special requirements.

Usage Guide

Storage:

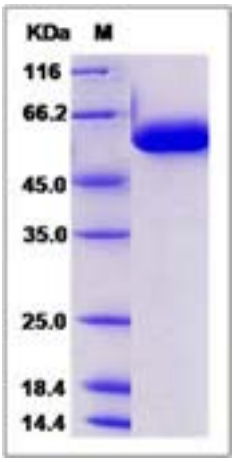
Store it under sterile conditions at -20℃ to -80℃ upon receiving. Recommend to aliquot the protein into smaller quantities for optimal storage.

Avoid repeated freeze-thaw cycles.

Reconstitution:

Detailed reconstitution instructions are sent along with the products.

SDS-PAGE:



Protein Description

FUCA1 is a lysosomal enzyme involved in the degradation of fucose-containing glycoproteins and glycolipids. Mutations in FUCA1 gene are associated with fucosidosis (FUCA1D), which is an autosomal recessive lysosomal storage disease. Different phenotypes include clinical features such as neurologic deterioration, growth retardation, visceromegaly, and seizures in a severe early form; coarse facial features, angiokeratoma corporis diffusum, spasticity and delayed psychomotor development in a longer surviving form; and an unusual spondylometaphyseoepiphyseal dysplasia in yet another form.

References

1.Yang M, *et al.* (1993) A mutation generating a stop codon in the alpha-L-fucosidase gene of a fucosidosis patient. Biochem Biophys Res Commun. 189(2):1063-8. 2.Fukushima H, *et al.* (1991) Sequencing and expression of a full-length cDNA for human alpha-L-fucosidase. J Inherit Metab Dis. 13(5):761-5. 3.Kretz KA, *et al.* (1990) Characterization of EcoRI mutation in fucosidosis patients: a stop codon in the open reading frame. J Mol Neurosci. 1(3):177-80.