Human ARL6IP6 Protein (Fc Tag)

Catalog Number: **14613-H04H**

General Information

Gene Name Synonym:

AIP-6; PFAAP1

Protein Construction:

A DNA sequence encoding the human ARL6IP6 (NP_689735.1) (Met1-Ser110) was expressed with the Fc region of mouse IgG1 at the N-terminus.

Source: Human

Expression Host: HEK293 Cells

QC Testing

Purity: > 95 % as determined by SDS-PAGE

Endotoxin:

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

Predicted N terminal: Asp

Molecular Mass:

The recombinant human ARL6IP6 consists 346 amino acids and predicts a molecular mass of 38.6 kDa.

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

Stability & Storage:
Samples are stable for twelve months from date of receipt at -20℃ to -80℃.

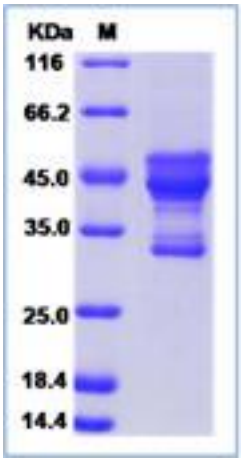
Store it under sterile conditions at -20℃ to -80℃ upon receiving. Recommend to aliquot the protein into smaller quantities for optimal storage.

Avoid repeated freeze-thaw cycles.

Reconstitution:

Detailed reconstitution instructions are sent along with the products.

SDS-PAGE:



Protein Description

It had been found that a homozygous truncating mutation in ARL6IP6 as the likely cause of a syndromic form of CMTC associated with major dysmorphism, developmental delay, transient ischemic attacks and cerebral vascular malformations. This gene was previously implicated by genome wide association study (GWAS) as a susceptibility locus to ischemic stroke in young adults. We identify ARL6IP6 as a novel candidate gene for a syndromic form of CMTC. This suggests that ischemic stroke or transient ischemic attacks (TIA) may represent, at least in some cases, the mild end of a phenotypic spectrum that has at its severe end autosomal recessive CMTC. This finding contributes to a growing appreciation of the continuum of Mendelian and common complex diseases.