

## ARL6IP6 Protein, Human, Recombinant (mFc)

### General Information

Synonyms:	PFAAP1;AIP-6;ADP-ribosylation factor-like 6 interacting protein 6
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. Predicted N terminal: Asp
Species:	Human
Expression Host:	HEK293 Cells
Accession:	Q8N6S5
Molecular Weight:	38.6 kDa (predicted)

### QC Testing

Biological Activity:	Activity testing is in progress. It is theoretically active, but we cannot guarantee it. If you require protein activity, we recommend choosing the eukaryotic expression version first.
Purity:	> 95 % as determined by SDS-PAGE
Endotoxin:	< 1.0 EU/μg of the protein as determined by the LAL method.
Formulation:	Lyophilized from a solution filtered through a 0.22 μm filter, containing PBS, pH 7.4. Typically, a mixture containing 5% to 8% trehalose, mannitol, and 0.01% Tween 80 is incorporated as a protective agent before lyophilization.

### Preparation and Storage

Reconstitution:	A Certificate of Analysis (CoA) containing reconstitution instructions is included with the products. Please refer to the CoA for detailed information.
Stability & Storage:	It is recommended to store recombinant proteins at -20°C to -80°C for future use. Lyophilized powders can be stably stored for over 12 months, while liquid products can be stored for 6-12 months at -80°C. For reconstituted protein solutions, the solution can be stored at -20°C to -80°C for at least 3 months. Please avoid multiple freeze-thaw cycles and store products in aliquots.
Shipping:	In general, Lyophilized powders are shipping with blue ice.

### Protein Background

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.

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