

Recombinant Human AIPL1 Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. AIPL1-2327H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	AIPL1 MS Standard C13 and N15-labeled recombinant protein (NP_055151) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
Species	Human
Source	HEK293
Description	Leber congenital amaurosis (LCA) is the most severe inherited retinopathy with the earliest age of onset and accounts for at least 5% of all inherited retinal diseases. Affected individuals are diagnosed at birth or in the first few months of life with nystagmus, severely impaired vision or blindness and an abnormal or flat electroretinogram. The photoreceptor/pineal-expressed gene, AIPL1, encoding aryl-hydrocarbon interacting protein-like 1, is located within the LCA4 candidate region. The encoded protein contains three tetratricopeptide motifs, consistent with chaperone or nuclear transport activity. Mutations in this gene may cause approximately 20% of recessive LCA. Alternative splicing results in multiple transcript variants.
Molecular Mass	43.9 kDa
AA Sequence	MDAALLNVEGVKKILHGGTGELPNFITGSRVIFHFRTMKCDEERTVIDDSRQVGQ PMHIIIGNMFKLEVWEILLTSMRVHEVAEFWCDTIHTGVYPILSRSLRQMAQGKDPT WHVHTCGLANMFAYHTLGIEDLDELQKEPQPLVFVIELLQVDAPSDYQRETWNLSN

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HEKMKAVPVLHGEGNRLFKLGRYEEASSKYQEAIICLRNLQTKEKPWEVQWLKLEK
MINTLILNYCQCLLKKEEYEVLEHTSDILRHHPGIVKAYYVRARAHAEVWNEAEAKA
DLQKVLELEPSMQKAVRRELRLLENRMAEKQEEERLRCRNMLSQGATQPPAEPPT
EPPAQSSTEPPAEPPTAPSAELSAGPPAEPATEPPPSPGHSLQHTRTRPLEQKLISE
EDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining

Stability Stable for 3 months from receipt of products under proper storage and handling conditions.

Storage Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

Concentration 50 µg/mL as determined by BCA

Storage Buffer 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name [AIPL1 aryl hydrocarbon receptor interacting protein-like 1 \[Homo sapiens \(human\) \]](#)

Official Symbol [AIPL1](#)

Synonyms AIPL1; aryl hydrocarbon receptor interacting protein-like 1; aryl hydrocarbon receptor interacting protein like 1, LCA4; aryl-hydrocarbon-interacting protein-like 1; LCA4; AIPL2;

Gene ID [23746](#)

mRNA Refseq [NM_014336](#)

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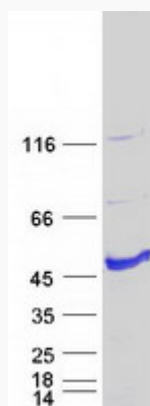
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Protein Refseq NP_055151

MIM 604392

UniProt ID Q9NZN9

SDS-PAGE



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