

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

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

## SPECIFICATION

<b>Product Overview</b>	AIPL1 MS Standard C13 and N15-labeled recombinant protein (NP_055151) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
<b>Species</b>	Human
<b>Source</b>	HEK293
<b>Description</b>	<p>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.</p>
<b>Molecular Mass</b>	43.9 kDa
<b>AA Sequence</b>	MDAALLNVEGVKKTILHGGTGELPNFITGSRVIFHFRTMKCDEERTVIDDSRQVGQ PMHIIIGNMFKLEVWEILLTSMRVHEVAEFWCDTIHTGVYPILSRSLRQMAQGKDPT WHVHTCGLANMFAYHTLGYEDLDELQKEPQPLVFVIELLQVDAPSDYQRETWNLSN

 Tel: 1-631-559-9269    1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)     Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

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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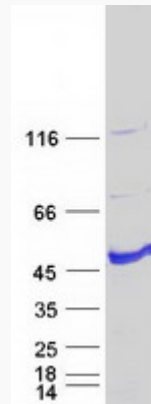
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**Protein Refseq** NP\_055151

**MIM** 604392

**UniProt ID** Q9NZN9

**SDS-PAGE**



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