

## Recombinant Human AIPL1, His-tagged

**Cat. No.** AIPL1-9509H    **Lot. No.** (See product label)

### SPECIFICATION

<b>Product Overview</b>	Recombinant Human AIPL1 protein, fused to His-tag, was expressed in E.coli and purified by Ni-sepharose.
<b>Species</b>	Human
<b>Source</b>	E.coli
<b>ProteinLength</b>	C-term-355a.a.
<b>Description</b>	<p>Leber congenital amaurosis (LCA) accounts for at least 5% of all inherited retinal disease and is the most severe inherited retinopathy with the earliest age of onset. Individuals affected with LCA are diagnosed at birth or in the first few months of life with severely impaired vision or blindness, nystagmus and an abnormal or flat electroretinogram. The photoreceptor/pineal -expressed gene, AIPL1, encoding aryl-hydrocarbon interacting protein-like 1, was mapped within the LCA4 candidate region. The protein contains three tetratricopeptide motifs, consistent with nuclear transport or chaperone activity. AIPL1 mutations may cause approximately 20% of recessive LCA.</p>
<b>Storage</b>	The protein is stored in PBS buffer at -20°C. Avoid repeated freezing and thawing cycles.
<b>Storage Buffer</b>	1M PBS (58mM Na <sub>2</sub> HPO <sub>4</sub> , 17mM NaH <sub>2</sub> PO <sub>4</sub> , 68mM NaCl, pH8. ) added with 300mM Imidazole and 0.7% Sarcosyl, 15%glycerol.

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## GENE INFORMATION

<b>Gene Name</b>	AIPL1 aryl hydrocarbon receptor interacting protein-like 1 [ Homo sapiens ]
<b>Official Symbol</b>	AIPL1
<b>Synonyms</b>	AIPL1; aryl hydrocarbon receptor interacting protein-like 1; aryl hydrocarbon receptor interacting protein like 1 , LCA4; aryl-hydrocarbon-interacting protein-like 1; LCA4; AIPL2;
<b>Gene ID</b>	23746
<b>mRNA Refseq</b>	NM_001033054
<b>Protein Refseq</b>	NP_001028226
<b>MIM</b>	604392
<b>UniProt ID</b>	Q9NZN9
<b>Chromosome Location</b>	17p13.1
<b>Function</b>	binding; farnesylated protein binding; unfolded protein binding;

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