

## Recombinant Human AIPL1 Protein, GST-tagged

**Cat. No.** AIPL1-480H    **Lot. No.** (See product label)

### SPECIFICATION

<b>Product Overview</b>	Human AIPL1 full-length ORF ( AAH12055, 1 a.a. - 384 a.a.) recombinant protein with GST-tag at N-terminal.
<b>Species</b>	Human
<b>Source</b>	Wheat Germ
<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. [provided by RefSeq, Jan 2014]</p>
<b>Molecular Mass</b>	67.98 kDa
<b>AA Sequence</b>	<p>MDAALLLNVEGVKKTILHGGTGELPNFITGSRVIFHFRTMKCDEERTVIDDSRQVGQ          PMHIIIGNMFKLEVWEILLTSMRVHEVAEFWCDTIHTGVYPILSRSLRQMAQGKDPT          WHVHTCGLANMFAYHTLGYEDLDELQKEPQPLVFVIELLQVDAPSDYQRETWNLSN          HEKMKAVPVLHGEGNRLFKLGRYEEASSKYQEAIICLRNLQTKEKPWEVQWLKLEK</p>

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MINTLILNYCQCLLKKEEYVEVLEHTSDILRHHPGIVKAYYVRARAHAEVWNEAEAKA  
DLQKVLELEPSMQKAVRRELRLLENRMAEKQEEERLRCRNMLSQGATQPPAEPPT  
EPPAQSSTEPPAEPPTAPSAELSAGPPAEPATEPPPPSPGHSLQH

<b>Applications</b>	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
<b>Notes</b>	Best use within three months from the date of receipt of this protein.
<b>Storage</b>	Store at -80centigrade. Aliquot to avoid repeated freezing and thawing.
<b>Storage Buffer</b>	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

## GENE INFORMATION

<b>Gene Name</b>	<a href="#">AIPL1 aryl hydrocarbon receptor interacting protein-like 1 [ Homo sapiens ]</a>
<b>Official Symbol</b>	<a href="#">AIPL1</a>
<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>	<a href="#">23746</a>
<b>mRNA Refseq</b>	<a href="#">NM_001033054</a>
<b>Protein Refseq</b>	<a href="#">NP_001028226</a>

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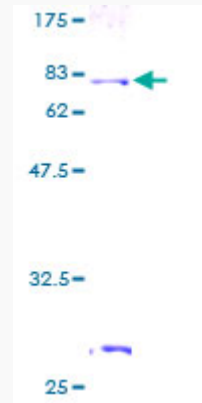
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**MIM** 604392

**UniProt ID** Q9NZN9

**Quality Control  
Testing:**



12.5% SDS-PAGE Stained with Coomassie Blue.

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