

Recombinant Full Length Human AIPL1 Protein, GST-tagged

Cat. No. AIPL1-960HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human AIPL1 full-length ORF (AAH12055, 1 a.a. - 384 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	384 amino acids
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. [provided by RefSeq, Jan 2014]
Molecular Mass	67.98 kDa
AA Sequence	MDAALLLNVE GVKKTILHGG TGELPNFITG SRVIFHFRTM KCDEERTVID DSRQVGQPMH IIIGNMFKLE VWEILLTSMR VHEVAEFWCD TIHTGVYPIL SRSLRQMAQG KDPTTEWHVHT CGLANMFAYH TLGYEDLDEL QKEPQPLVFV

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

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

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

IELLQVDAPS DYQRETWNLS NHEKMKAVPV LHGEGNRLFK LGRYEEASSK
 YQEAIICLRN LQTKEKPWEV QWLKLEKMIN TLILNYCQCL LKKEEYYEVL
 EHTSDILRHH PGIVKAYYVR ARAHAEVWNE AEAADLQKV LELEPSMQKA
 VRRELRLLEN RMAEKQEEER LRCRNMLSQG ATQPPAEPPT EPPAQSSTEP
 PAEPPTAPSA ELSAGPPAEP ATEPPPPSPGH SLQH

Applications
 Enzyme-linked Immunoabsorbent Assay
 Western Blot (Recombinant protein)
 Antibody Production
 Protein Array

Storage Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.

Storage Buffer 50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

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

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_001033054](#)

Protein Refseq [NP_001028226](#)

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MIM	604392
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UniProt ID	Q9NZN9
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