

## Product datasheet for PH304079

### AIPL1 (NM\_014336) Human Mass Spec Standard

#### Product data:

Product Type:	Mass Spec Standards
Description:	AIPL1 MS Standard C13 and N15-labeled recombinant protein (NP_055151)
Species:	Human
Expression Host:	HEK293
Expression cDNA Clone or AA Sequence:	RC204079
Predicted MW:	43.9 kDa
Protein Sequence:	>RC204079 protein sequence Red=Cloning site Green=Tags(s)

MDAALLLNVEGVKKTILHGGTGELPNFITGSRVIFHFRTMKCDEERTVIDDSRQVGQPMHIIIGNMFKLE  
VWEILLTSMRVHEVAEFWCDTIHTGVYPILSRSLRQMAQGDPTTEWHVHTCGLANMFAYHTLGYEDLDEL  
QKEPQPLVFVIELLQVDAPSDYQRETWNLSNHEKMKAVPVLHGEGNRLFKLGRYEEASSKYQEAIICLRN  
LQTKEKPWEVQWLKLEKMINTLILNYCQCLLKKEEYVELEHTSDILRHHPGIVKAYVVRARAHAEVWNE  
AEAKADLQKVLELEPSMQKAVRRELRLLENRMAEKQEEERLCRNMLSQGATQPPAEPPTPEPPAQSSTEP  
PAEPPTAPSAELSAEPPAEPATEPPPPSPGHSLSQH

TRTRPLEQKLI SEEDLAANDILDYKDDDDKV

Tag:	C-Myc/DDK
Purity:	> 80% as determined by SDS-PAGE and Coomassie blue staining
Concentration:	>0.05 µg/µL as determined by microplate BCA method
Labeling Method:	Labeled with [U- <sup>13</sup> C <sub>6</sub> , <sup>15</sup> N <sub>4</sub> ]-L-Arginine and [U- <sup>13</sup> C <sub>6</sub> , <sup>15</sup> N <sub>2</sub> ]-L-Lysine
Buffer:	25 mM Tris-HCl, 100 mM glycine, pH 7.3
Storage:	Store at -80°C. Avoid repeated freeze-thaw cycles.
Stability:	Stable for 3 months from receipt of products under proper storage and handling conditions.
RefSeq:	<u><a href="#">NP_055151</a></u>
RefSeq Size:	2990
RefSeq ORF:	1152
Synonyms:	AIPL2; LCA4
Locus ID:	23746



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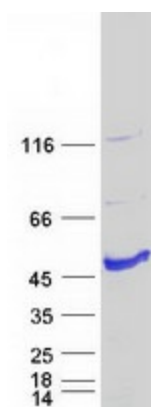
UniProt ID: [Q9NZN9](#), [F1T0B6](#)

Cytogenetics: 17p13.2

**Summary:** 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]

Protein Families: Druggable Genome

### Product images:



Coomassie blue staining of purified AIPL1 protein (Cat# [TP304079]). The protein was produced from HEK293T cells transfected with AIPL1 cDNA clone (Cat# [RC204079]) using MegaTran 2.0 (Cat# [TT210002]).