

## Product datasheet for **SC335698**

### AIPL1 (NM\_001285399) Human Untagged Clone

#### Product data:

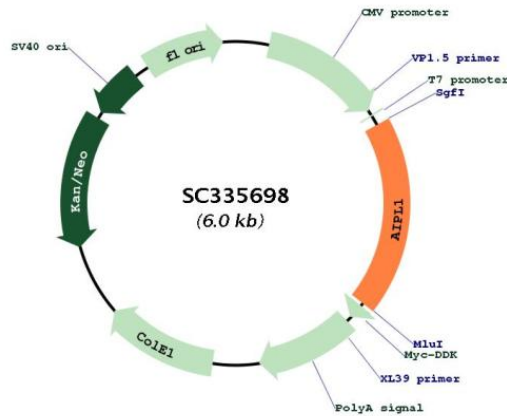
Product Type:	Expression Plasmids
Product Name:	AIPL1 (NM_001285399) Human Untagged Clone
Tag:	Tag Free
Symbol:	AIPL1
Synonyms:	AIPL2; LCA4
Mammalian Cell Selection:	Neomycin
Vector:	pCMV6-Entry (PS100001)
E. coli Selection:	Kanamycin (25 ug/mL)
Fully Sequenced ORF:	>SC335698 representing NM_001285399. Blue=Insert sequence Red=Cloning site Green=Tag(s)

GCTCGTTTAGTGAACCGTCAGAATTTTGTAAACGACTACTATAGGGCGCCGGGAATTCGTCGACTG  
GATCCGGTACCGAGGAGATCTGCCGCC**GCGATCGCC**  
ATGGATGCCGCTCTGCTCCTGAACGTGGAAGGGGTCAAGAAAACCATCTGCACGGGGGCACGGGCGAG  
CTCCAAACTTCATACCGGATCCCGAGAGCGGACAGTCATTGACGACAGTCGGCAGGTGGCCAGCCC  
ATGCACATCATCATCGAAACATGTTCAAGCTCGAGGTCTGGGAGATCCTGCTTACCTCCATGCCGGTG  
CACGAGGTGGCCGAGTTCTGGTGCACACCATCCACACGGGGTCTACCCATCCTATCCCGAGCCTG  
AGGAGATGGCCAGGGCAAGGACCCACAGAGTGGCACGTGCACACGTGCCGGCTGGCCAAATGTTT  
GCCTACCACACGCTGGGCTACGAGGACCTGGACGAGCTGCAGAAGGAGCCTCAGCCTCTGGTCTTTGTG  
ATCGAGCTGCTGCAGTTGATGCCCGAGTGATTACCAGAGGGAGACCTGGAACCTGAGCAATCATGAG  
AAGATGAAGGCGGTGCCCGTCTCCACGGAGAGGAAATCGGCTTTCAAGCTGGCCGCTACGAGGAG  
GCCTCTTCCAAGTACCAGGAGGCCATCATCTGCCTAAGGAACCTGCAGACCAAGGAGAAGCCATGGGAG  
GTGCAAGTGGTGAAGCTGGAGAAGATGATCAACTCTGATCCTCAACTACTGCCAGTGCCTGCTGAAG  
AAGGAGGAGTACTATGAGGTGCTGGAGCACACCAAGTATATTCTCCGGCACCCAGGCATCGTGAAG  
GCCTACTACGTGCGTGCCCGGCTCACGCAGAGGTGTGGAATGAGGCCGAGGCCAAGGCCGACCTCCAG  
AAAGTGTGGAGCTGGAGCCGTCATGCAGAAGGCGGTGCGCAGGGAGCTGAGGCTGCTGGAGAACCGC  
ATGGCGGAGAAGCAGGAGGAGCGGCTGCGCTGCCGGAACATGCTGAGCCAGGTGCCACGCAGCCT  
CCGCAGAGCCACCCACAGAGCCACCCGACAGTCATCCACAGAGCCACCTGCAGAGCCACCCACAGCA  
CCATCTGCAGAGCTGTCCGAGGGCCCCCTGCAGAGCCAGCCACAGAGCCACCCCGTCCCAGGGCAC  
TCGCTGCAGCACTGA  
**ACGCGT**ACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAATGATATCCTGGAT  
TACAAGGATGACGACGATAAGGTTTAAACGGCCGGC

Restriction Sites: SgfI-MluI



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**Plasmid Map:**


**ACCN:** NM\_001285399

**Insert Size:** 1119 bp

**OTI Disclaimer:** Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).

**Components:** The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

**Reconstitution Method:**

1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

**RefSeq:** [NM\\_001285399.2](#)

**RefSeq Size:** 2954 bp

**RefSeq ORF:** 1119 bp

**Locus ID:** 23746

**UniProt ID:** [Q9NZN9](#)

**Cytogenetics:** 17p13.2

**Protein Families:** Druggable Genome

**MW:** 42.4 kDa

**Gene 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]

Transcript Variant: This variant (4) uses an alternate splice site in the 5' coding region, compared to variant 1. This results in a shorter protein (isoform 4), compared to isoform 1.