

Product datasheet for **SC302690**

AIPL1 (NM_001033055) Human Untagged Clone

Product data:

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| Product Type: | Expression Plasmids |
| Product Name: | AIPL1 (NM_001033055) Human Untagged Clone |
| Tag: | Tag Free |
| Symbol: | AIPL1 |
| Synonyms: | AIPL2; LCA4 |
| Vector: | <u>pCMV6 series</u> |
| Fully Sequenced ORF: | >NCBI ORF sequence for NM_001033055, the custom clone sequence may differ by one or more nucleotides ATGGATGCCGCTCTGCTCCTGAACGTGGAAGGGGTCAAGAAAACCATCTGCACGGGGG ACGGGCGAGCTCCAAACTTCATCACCGGATCCCGACACACGGGGGTCTACCCCATCCTA TCCCGGAGCTGAGGCAGATGGCCAGGGCAAGGACCCACAGAGTGGCAGTGCACACG TGCGGGCTGGCCAACATGTTGCTACCACACGCTGGGCTACGAGGACCTGGACGAGCTG CAGAAGGAGCCTCAGCCTCTGGTCTTTGTGATCGAGCTGCTGCAGTTGATGCCCGAGT GATTACCAGAGGGAGACCTGGAACCTGAGCAATCATGAGAAGATGAAGGGGTGCCCGTC CTCCACGAGAGGGAAATCGGCTCTTCAAGCTGGGCCGCTACGAGGAGGCCTCTTCCAAG TACCAGGAGGCCATCATCTGCCTAAGGAACCTGCAGACCAAGGAGAAGCCATGGGAGGTG CAGTGGCTGAAGCTGGAGAAGATGATCAATACTCTGATCCTCACTACTGCCAGTGCCTG CTGAAGAAGGAGGAGTACTATGAGGTGCTGGAGCACACCAAGTATATTCTCCGGCACCAC CCAGGCATCGTGAAGGCTACTACGTGCGTGCCTGGGCTCACGCAGAGGTGTGGAATGAG GCCGAGGCCAAGGCGGACCTCCAGAAAGTGTGGAGCTGGAGCCGTCCATGCAGAAGGCG GTGCGCAGGGAGCTGAGGCTGCTGGAGAACCAGTGGCGGAGAAGCAGGAGGAGGAGCGG CTGCGCTGCCGGAACATGCTGAGCCAGGGTGCCACGCAGCCTCCCGCAGAGCCACCCACA GAGCCACCCGCACAGTCATCCACAGAGCCACCTGCAGAGCCACCCACAGCACCATCTGCA GAGCTGTCCGCAGGGCCCTGCAGAGCCAGCCACAGAGCCACCCCGTCCCGCAGGGCAC TCGCTGCAGCACTGA |
| Restriction Sites: | Please inquire |
| ACCN: | NM_001033055 |
| 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). |
| OTI Annotation: | This TrueClone is provided through our Custom Cloning Process that includes sub-cloning into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA. |



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| 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: | <ol style="list-style-type: none">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: | <u>NM_001033055.1, NP_001028227.1</u> |
| RefSeq Size: | 2801 bp |
| RefSeq ORF: | 975 bp |
| Locus ID: | 23746 |
| UniProt ID: | <u>Q9NZN9</u> |
| Cytogenetics: | 17p13.2 |
| Protein Families: | Druggable Genome |
| Gene Summary: | <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> <p>Transcript Variant: This variant (3) lacks an alternate in-frame exon in the 5' coding region, compared to variant 1. It encodes isoform 3, which is shorter than isoform 1.</p> |