

Product datasheet for SC312514

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C9orf72 (AK057806) Human Untagged Clone

Product data:

Product Type: Expression Plasmids

Product Name: C9orf72 (AK057806) Human Untagged Clone

Tag: Tag Free Symbol: C9orf72

Synonyms: ALSFTD; FTDALS; FTDALS1

Vector: <u>pCMV6 series</u>

Fully Sequenced ORF: >NCBI ORF sequence for AK057806, the custom clone sequence may differ by one or more

nucleotides

CTCAAG

Restriction Sites: Please inquire

ACCN: AK057806

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.

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: <u>AK057806.1</u>, <u>BAB71583.1</u>

RefSeq Size: 1564 bp
RefSeq ORF: 669 bp
Locus ID: 203228
Cytogenetics: 9p21.2

Gene Summary: The protein encoded by this gene plays an important role in the regulation of endosomal

trafficking, and has been shown to interact with Rab proteins that are involved in autophagy and endocytic transport. Expansion of a GGGGCC repeat from 2-22 copies to 700-1600 copies in the intronic sequence between alternate 5' exons in transcripts from this gene is associated with 9p-linked ALS (amyotrophic lateral sclerosis) and FTD (frontotemporal dementia) (PMID: 21944778, 21944779). Studies suggest that hexanucleotide expansions could result in the selective stabilization of repeat-containing pre-mRNA, and the accumulation of insoluble dipeptide repeat protein aggregates that could be pathogenic in FTD-ALS patients (PMID: 23393093). Alternative splicing results in multiple transcript variants encoding different

isoforms. [provided by RefSeq, Jul 2016]