

Product datasheet for **SC330353**

C9orf72 (NM_001256054) Human Untagged Clone

Product data:

Product Type: Expression Plasmids
Product Name: C9orf72 (NM_001256054) Human Untagged Clone
Tag: Tag Free
Symbol: C9orf72
Synonyms: ALSFTD; DENND9; DENNL72; FTDALS; FTDALS1
Vector: pCMV6-Entry (PS100001)
Fully Sequenced ORF: >SC330353 representing NM_001256054.
 Blue=Insert sequence Red=Cloning site Green=Tag(s)

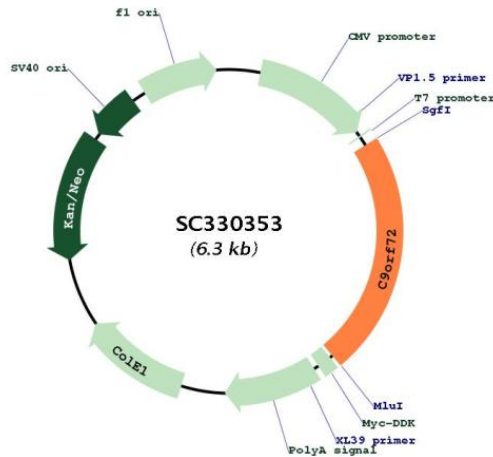
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Restriction Sites: Sgfl-Mlul



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Plasmid Map:


ACCN: NM_001256054

Insert Size: 1446 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_001256054.1](#)

RefSeq Size: 3339 bp

RefSeq ORF: 1446 bp

Locus ID: 203228

UniProt ID: [Q96LT7](#)

Cytogenetics: 9p21.2

MW: 54.3 kDa

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]

Transcript Variant: This variant (3) represents the longest transcript and encodes the longer isoform (a). Hexanucleotide expansions that can occur in a repeat region between the first and second exon of this variant have been associated with frontotemporal dementia and amyotrophic lateral sclerosis (FTD-ALS; PMID: 21944778, PMID: 21944779). Variants 2 and 3 encode the same protein.