

Product datasheet for SC326703

OriGene Technologies, Inc.

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Twinkle (TWNK) (NM_001163814) Human Untagged Clone

Product data:

Product Type: Expression Plasmids

Product Name: Twinkle (TWNK) (NM_001163814) Human Untagged Clone

Tag: Tag Free
Symbol: Twinkle

Synonyms: ATXN8; C10orf2; IOSCA; MTDPS7; PEO; PEO1; PEOA3; PRLTS5; SANDO; SCA8; TWINL

Mammalian Cell

Selection:

None

Vector: pCMV6-XL5

E. coli Selection: Ampicillin (100 ug/mL)

Fully Sequenced ORF: >NCBI ORF sequence for NM_001163814, the custom clone sequence may differ by one or

more nucleotides

GGCTCAGCCAAAGTGAGTGGCCTT

Restriction Sites: Please inquire ACCN: NM_001163814

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>NM 001163814.1</u>, <u>NP 001157286.1</u>

 RefSeq Size:
 1844 bp

 RefSeq ORF:
 387 bp

 Locus ID:
 56652

 Cytogenetics:
 10q24.31

Protein Families: Druggable Genome

Gene Summary: This gene encodes a hexameric DNA helicase which unwinds short stretches of double-

stranded DNA in the 5' to 3' direction and, along with mitochondrial single-stranded DNA binding protein and mtDNA polymerase gamma, is thought to play a key role in mtDNA replication. The protein localizes to the mitochondrial matrix and mitochondrial nucleoids. Mutations in this gene cause infantile onset spinocerebellar ataxia (IOSCA) and progressive external ophthalmoplegia (PEO) and are also associated with several mitochondrial depletion syndromes. Alternative splicing results in multiple transcript variants encoding distinct

isoforms.[provided by RefSeq, Aug 2009]

Transcript Variant: This variant (4) has multiple splice site differences, compared to variant 1.

The encoded protein (isoform D) lacks most of the N-terminus, which contains a

mitochondrial targeting sequence and probable ssDNA binding domain, and has a shorter and distinct C-terminus, which lacks a C-terminal domain required for hexamer formation,

compared to isoform A.