

Product datasheet for SC206229

OriGene Technologies, Inc.

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Nephronophthisis (NPHP1) (NM_000272) Human 3' UTR Clone

Product data:

Product Type: 3' UTR Clones

Product Name: Nephronophthisis (NPHP1) (NM_000272) Human 3' UTR Clone

Symbol: Nephronophthisis
Synonyms: JBTS4; NPH1; SLSN1

Mammalian Cell

Selection:

Neomycin

Vector: pMirTarget (PS100062)

ACCN: NM_000272

Insert Size: 473 bp

Insert Sequence: >SC206229 3'UTR clone of NM_000272

The sequence shown below is from the reference sequence of NM_000272. The complete

sequence of this clone may contain minor differences, such as SNPs.

Blue=Stop Codon Red=Cloning site

GGCAAGTTGGACGCCCGCAAGATCCGCGAGATTCTCATTAAGGCCAAGAAGGGCGGAAAGATCGCCGTG

TAACAATTGGCAGAGCTCAGAATTCAAGCGATCGCC

TCTAGTATCCAAAATATTTAAATTTATGAGTGTTAATAAAATTTATCTTGTTCAATGAA

CGAGATTTCGATTCCACCGCCGCCTTCTATGAAAGG

Restriction Sites: Sgfl-Mlul

OTI Disclaimer: Our molecular clone sequence data has been matched to the sequence identifier above as a

point of reference. Note that the complete sequence of this clone is largely the same as the

reference sequence but may contain minor differences, e.g., single nucleotide

polymorphisms (SNPs).

Components: The cDNA clone is shipped in a 2-D bar-coded Matrix tube as 10 ug dried plasmid DNA. The

package also includes 100 pmols of both the corresponding 5' and 3' vector primers in

separate vials.





RefSeq: <u>NM 000272.5</u>

Summary: This gene encodes a protein with src homology domain 3 (SH3) patterns. This protein

interacts with Crk-associated substrate, and it appears to function in the control of cell division, as well as in cell-cell and cell-matrix adhesion signaling, likely as part of a

multifunctional complex localized in actin- and microtubule-based structures. Mutations in this gene cause familial juvenile nephronophthisis type 1, a kidney disorder involving both tubules and glomeruli. Defects in this gene are also associated with Senior-Loken syndrome type 1, also referred to as juvenile nephronophthisis with Leber amaurosis, which is

characterized by kidney and eye disease, and with Joubert syndrome type 4, which is characterized by cerebellar ataxia, oculomotor apraxia, psychomotor delay and neonatal breathing abnormalities, sometimes including retinal dystrophy and renal disease. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by

RefSeq, Jul 2008]

Locus ID: 4867 **MW:** 18.8