

Product datasheet for RC226024L4V

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Nephronophthisis (NPHP1) (NM_001128179) Human Tagged ORF Clone Lentiviral Particle

Product data:

Product Type: Lentiviral Particles

Symbol: Nephronophthisis

Synonyms: JBTS4; NPH1; SLSN1

Mammalian Cell Puromycin

Selection:

Vector: pLenti-C-mGFP-P2A-Puro (PS100093)

Tag: mGFP

ACCN: NM_001128179

ORF Size: 1842 bp

ORF Nucleotide Sequence: The ORF insert of this clone is exactly the same as(RC226024).

OTI Disclaimer: The molecular sequence of this clone aligns with the gene accession number as a point of

reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing

variants is recommended prior to use. More info

OTI Annotation: This clone was engineered to express the complete ORF with an expression tag. Expression

varies depending on the nature of the gene.

RefSeq: <u>NM_001128179.1</u>

RefSeq ORF: 1845 bp

Locus ID: 4867

UniProt ID: O15259

Cytogenetics: 2q13

Protein Families: Druggable Genome





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MW:

69.7 kDa

Gene 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]