

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:	NM_001009944.3
Summary:	This gene encodes a member of the polycystin protein family. The encoded glycoprotein contains a large N-terminal extracellular region, multiple transmembrane domains and a cytoplasmic C-tail. It is an integral membrane protein that functions as a regulator of calcium permeable cation channels and intracellular calcium homeostasis. It is also involved in cell-cell/matrix interactions and may modulate G-protein-coupled signal-transduction pathways. It plays a role in renal tubular development, and mutations in this gene cause autosomal dominant polycystic kidney disease type 1 (ADPKD1). ADPKD1 is characterized by the growth of fluid-filled cysts that replace normal renal tissue and result in end-stage renal failure. Splice variants encoding different isoforms have been noted for this gene. Also, six pseudogenes, closely linked in a known duplicated region on chromosome 16p, have been described. [provided by RefSeq, Oct 2008]
Locus ID:	5310
MW:	37.1