

Recombinant Human NPHS1

Cat. No. NPHS1-29301TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant fragment corresponding to amino acids 33-122 of Human Nephlin with an N terminal proprietary tag: Predicted MWt 35.53 kDa.
Species	Human
Source	Wheat Germ
ProteinLength	90 amino acids
Description	This gene encodes a member of the immunoglobulin family of cell adhesion molecules that functions in the glomerular filtration barrier in the kidney. The gene is primarily expressed in renal tissues, and the protein is a type-1 transmembrane protein found at the slit diaphragm of glomerular podocytes. The slit diaphragm is thought to function as an ultrafilter to exclude albumin and other plasma macromolecules in the formation of urine. Mutations in this gene result in Finnish-type congenital nephrosis 1, characterized by severe proteinuria and loss of the slit diaphragm and foot processes.
Molecular Weight	35.530kDa inclusive of tags
Tissue specificity	Specifically expressed in podocytes of kidney glomeruli.
Form	Liquid
Purity	Proprietary Purification

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Storage buffer	pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.
Sequences of amino acids	GFWALPENLTVVEGASVELRCGVSTPGSAVQWAKDGLLLGPDPRIPGFPRYRLEGD PARGEFHLHIEACDLSDDAEYECQVGRSEMGPEL
Sequence Similarities	Belongs to the immunoglobulin superfamily.Contains 1 fibronectin type-III domain.Contains 8 Ig-like C2-type (immunoglobulin-like) domains.

GENE INFORMATION

Gene Name	NPHS1 nephrosis 1, congenital, Finnish type (nephrin) [Homo sapiens]
Official Symbol	NPHS1
Synonyms	NPHS1; nephrosis 1, congenital, Finnish type (nephrin); nephrin; CNF; NPHN;
Gene ID	4868
mRNA Refseq	NM_004646
Protein Refseq	NP_004637
MIM	602716
Uniprot ID	O60500
Chromosome Location	19q12-q13.1

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Pathway	Cell-Cell communication, organism-specific biosystem; Nephrin interactions, organism-specific biosystem; Nephrin/Neph1 signaling in the kidney podocyte, organism-specific biosystem;
Function	alpha-actinin binding; protein binding; protein domain specific binding; spectrin binding;

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