

# Recombinant Human NPHS1

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

## SPECIFICATION

|                           |  |
|---------------------------|--|
| <b>Product Overview</b>   | Recombinant fragment corresponding to amino acids 33-122 of Human Nephtrin with an N terminal proprietary tag: Predicted MWt 35.53 kDa.  |
| <b>Species</b>            | Human  |
| <b>Source</b>             | Wheat Germ   |
| <b>ProteinLength</b>      | 90 amino acids   |
| <b>Description</b>        | <p>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.</p> |
| <b>Molecular Weight</b>   | 35.530kDa inclusive of tags  |
| <b>Tissue specificity</b> | Specifically expressed in podocytes of kidney glomeruli.   |
| <b>Form</b>               | Liquid   |
| <b>Purity</b>             | Proprietary Purification   |

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

## GENE INFORMATION

|                            |  |
|----------------------------|--|
| <b>Gene Name</b>           | <a href="#">NPHS1 nephrosis 1, congenital, Finnish type (nephrin) [ Homo sapiens ]</a> |
| <b>Official Symbol</b>     | <a href="#">NPHS1</a>  |
| <b>Synonyms</b>            | NPHS1; nephrosis 1, congenital, Finnish type (nephrin); nephrin; CNF; NPHN;            |
| <b>Gene ID</b>             | <a href="#">4868</a>   |
| <b>mRNA Refseq</b>         | <a href="#">NM_004646</a>  |
| <b>Protein Refseq</b>      | <a href="#">NP_004637</a>  |
| <b>MIM</b>                 | <a href="#">602716</a>   |
| <b>Uniprot ID</b>          | <a href="#">O60500</a>   |
| <b>Chromosome Location</b> | 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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