

Table 1: Major genetic mutations responsible for familial proteinuric renal diseases.

G e n e t i c locus	Protein coded	Protein function	Phenotype	Treatment
<i>ACTN4</i>	α -Actinin 4	Cell interactions	FSGS, MCD	Resistant to immunosuppression
<i>ARHGAP24</i>	Rho GTPase Activating Protein 24	Inhibits Rac1-dependent membrane ruffling and epithelial woundhealing.	FSGS	Resistant to immunosuppression
<i>CD2AP</i>	CD2 Associated Protein	Links nephrin and podocin to phosphoinositide 3-OH kinase	FSGS	Resistant to immunosuppression
<i>COL4A3</i>	Collagen, Type IV, Alpha 3	Structure of glomerular basal membrane	Alport syndrome	ACE inhibitors
<i>NPHS1</i>	Nephrin	Essential componenet of slit diaphragm	Finnish-type nephrotic syndrome	Resistant to immunosuppression
<i>NPHS2</i>	Podocin	Recruitment of nephrin to slit diaphragm	Familial FSGS	Resistant to immunosuppression
<i>COQ2</i>	Coenzyme Q2	Part of the CoQ10 pathway and encodes the para-hydroxybenzoate-polyprenyltransferase	SRNS	Coenzyme Q10
<i>LAMB2</i>	Laminin Subunit Beta 2	Major laminin component of the glomerular basemant membrane	Pierson syndrome	Resistant to immunosuppression
<i>TRPC6</i>	Transient Receptor Potential Cation Channel Subfamily C Member 6	Contribute to calcium entry in podocytes	Familial FSGS	Sildenafil
<i>ITGA3</i>	Integrin Subunit Alpha 3	Podocyte–GBM interaction	FSGS	Resistant to immunosuppression

FSGS: focal segmental glomerulosclerosis, GMB: glomerular basal membrane, MCD: minimal change disease, SRNS: steroid resistant nephrotic syndrome.