NPHS1

Nephrotic Syndrome

Mode of Inheritance	• **Autosomal recessive**
Renal Phenotype	• **Congenital nephrotic syndrome** • **Age of onset: typically within first 3 months of life** • **Typical biopsy findings: MCD, FSGS, DMS**

Extra-renal Manifestations	• **Sequelae from nephrotic syndrome:**
	  • **Hypoalbuminemia and edema**
	  • **Hyperlipidemia**
	  • **Hypothyroidism**
	  • **Loss of immunoglobulins**

Pre-Transplant Management	• **Avoidance of steroid and intensive immunosuppression therapy** • **Avoidance of renal biopsy**
Transplant Considerations	• **Tailor immunosuppression given lower risk of recurrence post-transplant**
Post-Transplant Management	• **In general, lower risk of disease recurrence (4.5% vs 28.5%) Trautmann *CJASN* 10:592, 2015)** • **Some reports of higher FSGS recurrence rate with the Fin(major) allele (c.121delCT) due to formation of anti-nephrin antibodies (Holmberg *Ped Nephrol* 29:2309, 2014)**

https://www.omim.org/entry/256300