AGXT
Primary Hyperoxaluria, type 1

Mode of Inheritance	• Autosomal recessive
Renal Phenotype	• Nephrolithiasis, nephrocalcinosis, renal failure • Age of onset: Variable, usually childhood but can also be in infancy or adulthood

Extra-renal Manifestations	• Systemic oxalate deposition, leading to:
	  • Optic atrophy and retinopathy
	  • Heart block
	  • Peripheral vascular disease
	  • Osteosclerosis, bone pain, and pathologic fractures
	  • Calcinosis cutis metastatica
	  • Peripheral neuropathy

Pre-Transplant Management	• Trial of pyridoxine • Frequent dialysis (as much as 5 or 6 days per week) to bring down oxalate levels • Priority listing for transplantation to minimize tissue oxalate deposition
Transplant Considerations	• Combined liver-kidney transplant is essential to address underlying metabolic defect
Post-Transplant Management	• Low risk of disease recurrence if combined liver-kidney transplant

https://www.omim.org/entry/259900