Web Table I Genetic Syndromes Associated with Renal Artery Stenosis in Children and Adolescents

Genetic condition	*Mutation*	*Inheritance*	*Clinical association(s)*	*Renal vascular malformation*
Neurofibromatosis 1	*NF1* gene	AD	Neurofibromas - Café au lait macules, optic glioma, increased risk of tumors - pheochromocytoma, Lisch nodules	RAS, External compression - Wilm tumor
Tuberous sclerosis	TSC1, TSC2	AD	Tubers (glial nodules), seizures, adenoma sebaceum, myocardial rhabdomyomas, shagreen patch, ash-leaf macules	RAS, MAS, renal angiomyolipoma
Turner's syndrome	XO	Non-disjunction	Streak gonads, primary amenorrhea, short stature, webbed neck	Coarctation of aorta, MAS, RAS
Marfan's syndrome	Fibrillin	AD	Arachnodactyly, dissecting aortic aneurysm, ectopic lens, mitral valve prolapse	Aortic aneurysm, renal aneurysm, RAS
Loeys-Dietz syndrome	TGFBR1, TGFBR2, SMD3, TGRB2, TGFB3	AD, sporadic	Aortic aneurysm, aortic dissection, craniosynostosis, pes planus, scoliosis, hypertelorism, bifid uvula	RAS, renal artery aneurysm, coarctation of aorta
Alagille syndrome	JAG1, NOTCH2	AD	Xanthomas, cholestatic liver disease, pulmonic stenosis, broad forehead, deep-set eyes	Coarctation of aorta, RAS
Williams-Beuren syndrome	Deletion of genes in chromosome 7	AD, sporadic	Broad forehead, wide mouth, supravalvular aortic stenosis, developmental delay	MAS, RAS, coarctation of aorta, hypoplasia of the aortic arch
Hereditary Nephropathy, Aneurysms, and Muscle Cramps	COL4A1	AD	Intracranial aneurysms, arterial retinal tortuosity, cataracts, muscle cramps	Cystic compression of renal vessels
Alport syndrome	COL4A3, COL4A4, COL4A5	X-linked (common), AD AR	Sensorineural hearing loss, anterior lenticonus, renal dysfunction	RAS
Idiopathic infantile arterial calcification	ENPP1, ABCC6	AR	Heart failure, respiratory distress, cyanosis	Vaso-occlusive RAS
Autosomal dominant polycystic kidney disease	PKD1, PKD2	AD	Recurrent UTI, kidney stones, heart valve abnormalities, aneurysms	Cystic compression of renal vessels, RAS
Autosomal recessive polycystic kidney disease	PKHD1	AR	Failure to thrive, respiratory failure, enlarged kidneys, oligohydramnios	Cystic compression of renal vessels, RAS

AD: Autosomal Dominant; AR: Autosomal Recessive; RAS: Renal artery stenosis; MAS: Mid-aortic syndrome.