Pathogenic/likely pathogenic variants in *COL4A3* are associated with *COL4A3*-related Alport syndrome. Single heterozygous pathogenic/likely pathogenic variants are associated with the autosomal dominant form of *COL4A3*-related Alport syndrome which is characterized by variable kidney involvement ranging from asymptomatic, isolated intermittent hematuria to chronic kidney disease that may or may not progress to kidney failure. Adult-onset hearing loss and ocular involvement may occur, warranting consideration of baseline evaluations at time of diagnosis. Biallelic pathogenic/likely pathogenic variants are associated with the autosomal recessive form of *COL4A3*-related Alport syndrome which is characterized by early onset of variable kidney abnormalities including microhematuria with or without proteinuria, thin basement membrane disease (TBMD), focal segmental glomerular sclerosis (FSGS), and kidney failure by early- to mid-adulthood. Early-onset sensorineural hearing loss (SNHL) and ocular involvement can include bilateral hearing loss, anterior lenticonus, maculopathy, corneal endothelial vesicles, and recurrent corneal erosion. MedGen UID: 1648334, 1648326.