

ID	Genes	Variants (ACMG classification)	Kidney phenotype	Extrarenal phenotype	Laboratory analysis	Ref.
III-7	SLC34A3 (het)	<i>c.1561dupC, p.Leu521Profs*72</i> (LP)	bilateral kidney stones	short stature, rickets	hypophosphatemia, hypercalciuria	[15]
	SLC34A1 (het)	<i>c.1535G>A, p.Arg512His</i> (VUS)				
III-6	SLC34A3 (het)	<i>c.1561dupC, p.Leu521Profs*72</i> (LP)	bilateral kidney stones	short stature, rickets	hypophosphatemia	[15]
	SLC34A1 (het)	<i>c.1535G>A, p.Arg512His</i> (VUS)				
II-5	SLC34A3 (het)	<i>c.1561dupC, p.Leu521Profs*72</i> (LP)	no	rickets, short stature	normal serum phosphate	[15]
	SLC34A1 (het)	<i>c.1535G>A, p.Arg512His</i> (VUS)				
2	SLC34A3 (het)	<i>c.496G>A, p.Gly166Ser</i> (P)	recurrent nephrolithiasis	no	hypophosphatemia, CKD G2	[16]
	SLC34A3 (het)	<i>c.1093+41_1094-15del, p.?</i> (LP)				
	SLC34A1 (het)	<i>c.272_292del, p.Val91_Ala97del</i> (VUS)				
4	SLC34A3 (het)	<i>c.496G>A, p.Gly166Ser</i> (P)	recurrent nephrolithiasis	short stature	hypophosphatemia, hypercalciuria, CKD G1	[16]
	SLC34A1 (het)	<i>c.1416+3G>A, p.?</i> (VUS)				
	NHERF1 (het)	<i>c.328C>G, p.Leu110Val</i> (LP)				
6	SLC34A3 (hom)	<i>c.575C>T, p.Ser192Leu</i> (LP)	recurrent nephrolithiasis	mineralization disorder	hypophosphatemia, CKD G2	[16]
	NHERF1 (het)	<i>c.328C>G, p.Leu110Val</i> (LP)				
8	SLC34A3 (het)	<i>c.781A>G, p.Ser261Gly</i> (LB)	mild	no	hypophosphatemia	[16]
	SLC34A1 (het)	<i>c.644+5G>A, p.?</i> (VUS)				

Supplementary Table S1: Previous reported case series of patients carrying digenic or oligogenic variants in genes associated with kidney stone disease due to renal phosphate wasting including their genotype and kidney/extrarenal phenotype.

Abbreviations: *het* heterozygous; *hom* homozygous; *LB* likely benign; *VUS* variant of unknown significance; *LP* likely pathogenic; *P* pathogenic; *CKD* chronic kidney disease; *CKD stages (G)* according KDIGO classification); *Ref.* Reference.