

SUPPLEMENTARY TABLE

for

“Dominant negative mutation in oxalate transporter *SLC26A6* associated with enteric hyperoxaluria and nephrolithiasis” by Nicolas Cornière, R. Brent Thomson, Stéphanie Thauvin, Bruno O. Villoutreix, Sophie Karp, Diane W. Dynia, Sarah Burlein, Lennart Brinkmann, Alaa Badreddine, Aurélie Dechaume, Mehdi Derhourhi, Emmanuelle Durand, Emmanuel Vaillant, Philippe Froguel, Régine Chambrey, Peter S. Aronson, Amélie Bonnefond, & Dominique Eladari

Supplementary Table 1. List of genes investigated in the present study

Gene ID	Gene name	NM ID	Location	Inheritance	Phenotype	Urolithiasis	Salt-losing nephropathy	Proximal tubulopathy
<i>ADCY10</i>	adenylate cyclase 10	NM_018417.5	1q24.2	candidate				
<i>AGXT</i>	alanine--glyoxylate and serine--pyruvate aminotransferase	NM_000030.2	2q37.3	AR	Hyperoxaluria	×		
<i>ALPL</i>	alkaline phosphatase, biomineralization associated	NM_000478.5	1p36.12	AD/AR	Hypophosphatasia	×		
<i>APRT</i>	adenine phosphoribosyltransferase	NM_000485.2	16q24.3	AR	Urolithiasis, Renal failure	×		
<i>AQP1</i>	aquaporin 1	NM_198098.3	7p14.3	candidate				
<i>ATP6V0A4</i>	ATPase H+ transporting V0 subunit a4	NM_020632.2	7q34	AR	Distal renal tubular acidosis	×		
<i>ATP6V1B1</i>	ATPase H+ transporting V1 subunit B1	NM_001692.3	2p13.3	AR	Renal tubular acidosis	×		
<i>BSND</i>	barttin CLCNK type accessory subunit beta	NM_057176.2	1p32.3	AR	Barter syndrome		×	
<i>CA2</i>	carbonic anhydrase 2	NM_000067.2	8q21.2	AR	Osteopetrosis with renal tubular acidosis	×	×	×
<i>CASR</i>	calcium sensing receptor	NM_000388.3	3q13.33-q21.1	AD/AR	Hypocalcemia, Barter syndrome	×	×	
<i>CFTR</i>	CF transmembrane conductance regulator	NM_000492.3	7q31.2	AR	Cystic fibrosis	×		
<i>CLCN5</i>	chloride voltage-gated channel 5	NM_000084.4	Xp11.23	XLR	Dent disease, Hypophosphatemic rickets, Nephrolithiasis, Proteinuria with hypercalciuric nephrocalcinosis	×		
<i>CLCNKB</i>	chloride voltage-gated channel Kb	NM_000085.4	1p36.13	AR	Barter syndrome	×	×	
<i>CLDN14</i>	claudin 14	NM_144492.2	21q22.13	candidate				
<i>CLDN16</i>	claudin 16	NM_006580.3	3q28	AR	Hypomagnesemia, Hypercalciuria, Nephrocalcinosis	×		

CLDN19	claudin 19	NM_148960.2	1p34.2	AR	Hypomagnesemia, Renal abnormalities	x		
CYP24A1	cytochrome P450 family 24 subfamily A member 1	NM_000782.4	20q13.2	AR	Infantile hypercalcemia	x		
DMP1	dentin matrix acidic phosphoprotein 1	NM_004407.3	4q22.1	AR	Hypophosphatemic rickets	x		
ENPP1	ectonucleotide pyrophosphatase/phosphodiesterase 1	NM_006208.2	6q23.2	AR	Hypophosphatemic rickets	x		
FAM20A	FAM20A golgi associated secretory pathway pseudokinase	NM_017565.3	17q24.2	AR	Enamel-renal syndrome, Nephrocalcinosis	x		
FGF23	fibroblast growth factor 23	NM_020638.2	12p13.32	AD/AR	Hypophosphatemic rickets	x		
GRHPR	glyoxylate and hydroxypyruvate reductase	NM_012203.1	9p13.2	AR	Hyperoxaluria	x		
HNF4A	hepatocyte nuclear factor 4 alpha	NM_175914.4	20q13.12	AD	Fanconi renotubular syndrome	x		x
HOGA1	4-hydroxy-2-oxoglutarate aldolase 1	NM_138413.3	10q24.2	AR	Hyperoxaluria	x		
HPRT1	hypoxanthine phosphoribosyltransferase 1	NM_000194.2	Xq26.2-q26.3	XLR	Lesch-Nyhan syndrome	x		
INVS	inversin	NM_014425.4	9q31.1	AR	Infantile nephronophthisis	x		
KCNJ1	potassium inwardly rectifying channel subfamily J member 1	NM_000220.4	11q24.3	AR	Barter syndrome	x	x	
NPHP1	nephrocystin 1	NM_000272.3	2q13	AR	Nephronophthisis	x		
NPHP3	nephrocystin 3	NM_153240.4	3q22.1	AD/AR	Nephronophthisis	x		
OCRL	OCRL inositol polyphosphate-5-phosphatase	NM_000276.3	Xq26.1	XLR	Dent disease, Lowe syndrome	x		
PHEX	phosphate regulating endopeptidase homolog X-linked	NM_000444.5	Xp22.11	XLD	Hypophosphatemic rickets	x		
PRPS1	phosphoribosyl pyrophosphate synthetase 1	NM_002764.3	Xq22.3	XLR	Gout, PRPS-related	x		
SLC12A1	solute carrier family 12 member 1	NM_000338.2	15q21.1	AR	Barter syndrome	x	x	
SLC22A12	solute carrier family 22 member 12	NM_144585.3	11q13.1	AR	Renal hypouricemia	x		
SLC26A1	solute carrier family 26 member 1	NM_213613.3	4p16.3	AR	Nephrolithiasis	x		
SLC26A6	solute carrier family 26 member 6	NM_022911.2	3p21.31	candidate				
SLC2A9	solute carrier family 2 member 9	NM_020041.2	4p16.1	AD/AR	Renal hypouricemia	x		
SLC34A1	solute carrier family 34 member 1	NM_003052.4	5q35.3	AD/AR	Fanconi renotubular syndrome, Infantile hypercalcemia, Hypophosphatemic nephrolithiasis and osteoporosis	x		
SLC34A3	solute carrier family 34 member 3	NM_080877.2	9q34.3	AR	Hypophosphatemic rickets, Hypercalciuria	x		
SLC3A1	solute carrier family 3 member 1	NM_000341.3	2p21	AR	Cystinuria	x		
SLC4A1	solute carrier family 4 member 1 (Diego blood group)	NM_000342.3	17q21.31	AD/AR	Distal renal tubular acidosis	x		
SLC7A9	solute carrier family 7 member 9	NM_014270.4	19q13.11	AD/AR	Cystinuria	x		
SLC9A3R1	SLC9A3 regulator 1	NM_004252.4	17q25.1	AD	Infantile Hypercalcemia, Hypophosphatemia, Nephrolithiasis	x		

<i>TRPV5</i>	transient receptor potential cation channel subfamily V member 5	NM_019841.6	7q34	candidate				
<i>TRPV6</i>	transient receptor potential cation channel subfamily V member 6	NM_018646.5	7q34	candidate				
<i>UMPS</i>	uridine monophosphate synthetase	NM_000373.3	3q21.2	AR	Oroticaciduria	×		
<i>VDR</i>	vitamin D receptor	NM_001017535.1	12q13.11	AR	Rickets	×		
<i>XDH</i>	xanthine dehydrogenase	NM_000379.3	2p23.1	AR	Xanthinuria	×		

AD, autosomal dominant; ***AR***, autosomal recessive; ***XLD***, X-linked dominant; ***XLR***, X-linked recessive