

Supplementary Table 1. Thirty genes that are known to cause monogenic forms of NL/NC and that were included in this study.

	Gene Symbol	Gene Name	Accession #	Disease entity	MIM-Phenotype #	Mode	Coding Exons	Reference
1	AGXT	Alanine-glyoxylate aminotransferase	NM_000030.2	Primary hyperoxaluria, type 1	259900	AR	11	1
2	APRT	Adenine phosphoribosyltransferase	NM_000485.2	Adenine phosphoribosyltransferase deficiency, Urolithiasis (DHA stones), renal failure	614723	AR	5	2
3	ATP6V0A4	ATPase, H+ transporting, lysosomal V0 subunit a4	NM_020632.2	dRTA	602722	AR	20	3
4	ATP6V1B1	ATPase, H+ transporting, lysosomal 56/58kDa, V1 subunit B1	NM_001692.3	Distal renal tubular acidosis (dRTA) with deafness	267300	AR	14	4
5	CA2	Carbonic anhydrase II	NM_000067.2	Osteopetrosis + d/pRTA	259730	AR	7	5
6	CASR	Calcium-sensing receptor	NM_001178065.1	Hypocalcemia with Bartter syndrome / hypocalcemia, autosomal dominant	601198	AD	6	6
7	CLCN5	Chloride channel, voltage-sensitive 5	NM_001127898.3	Dent disease / Nephrolithiasis, type 1	300009 / 310468	XR	14	7
8	CLCNKB	Chloride channel, voltage-sensitive Kb	NM_000085.4	Bartter syndrome, type 3	607364	AR	19	8
9	CLDN16	Claudin 16	NM_006580.3	Familial hypomagnesemia with hypercalciuria & nephrocalcinosis (FHHNC)	248250	AR	5	9
10	CLDN19	Claudin 19	NM_001123395.1	Familial hypomagnesemia with hypercalciuria & nephrocalcinosis (FHHNC) with ocular abnormalities	248190	AR	4	10
11	CYP24A1	Cytochrome P450, family 24, subfamily A, polypeptide 1	NM_000782.4	1,25-(OH) vitamin D-24 hydroxylase deficiency, infantile hypercalcemia	143880	AR	11	11
12	FAM20A	Family with sequence similarity 20, member A	NM_017565.3	Enamel-renal syndrome, amelogenesis imperfect and nephrocalcinosis	204690	AR	12	12
13	GRHPR	Glyoxylate reductase/hydroxypyruvate reductase	NM_012203.1	Primary hyperoxaluria, type 2	260000	AR	9	13
14	HNF4A	Hepatocyte nuclear factor 4, alpha	NM_000457.4	Mature onset diabetes of the young Fanconi syndrome + Nephrocalcinosis	125850	AD	1	14
15	HOGA1	4-hydroxy-2-oxoglutarate aldolase 1	NM_138413.3	Primary hyperoxaluria, type 3	613616	AR	7	15
16	HPRT1	Hypoxanthine phosphoribosyltransferase 1	NM_000194.2	Kelley-Seegmiller syndrome, partial HPRT deficiency, HPRT-related gout	300323	XR	9	16
17	KCNJ1	Potassium inwardly-rectifying channel, subfamily J, member 1	NM_000220.4	Bartter syndrome, type 2	241200	AR	2	17
18	OCRL	Oculocerebrorenal syndrome of Lowe	NM_000276.3	Lowe syndrome / Dent disease 2	309000 / 300555	XR	24	18

19	SLC12A1	Solute carrier family 12, member 1	NM_000338.2	Bartter syndrome, type 1	601678	AR	27	19
20	SLC22A12	Solute carrier family 22 (organic anion/urate transporter), member 12	NM_144585.3	Renal hypouricemia, RHUC1	220150	AR/AD	10	20
21	SLC26A1	Solute carrier family 26 (sulfate transporter), member 1	NM_213613	Nephrolithiasis, calcium oxalate	167030	AR	4	21
22	SLC2A9	Solute carrier family 2 (facilitated glucose transporter), member 9	NM_001001290.1	Renal hypouricemia, RHUC2	612076	AR/AD	13	22
23	SLC34A1	Solute carrier family 34 (sodium phosphate), member 1	NM_003052.4	Hypophosphatemic nephrolithiasis/osteoporosis-1, NPHLOP1 / Fanconi renotubular syndrome 2	612286 / 613388	AR/AD	13	23
24	SLC34A3	Solute carrier family 34 (sodium phosphate), member 3	NM_001177316.1	Hypophosphatemic rickets with hypercalciuria	241530	AR	12	24
25	SLC3A1	Solute carrier family 3 (cystine, dibasic and neutral amino acid transporters, activator of cystine, dibasic and neutral amino acid transport), member 1	NM_000341.3	Cystinuria, type A	220100	AR	10	25
26	SLC4A1	Solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group)	NM_000342.3	Primary distal renal tubular acidosis, dominant / recessive	179800 / 611590	AR/AD	19	26
27	SLC7A9	Solute carrier family 7 (glycoprotein associated amino acid transporter light chain, bo,+ system), member 9	NM_014270.4	Cystinuria, type B	220100	AR/AD	12	27
28	SLC9A3R1	Solute carrier family 9, subfamily A (NHE3, cation proton antiporter 3), member 3 regulator 1	NM_004252.4	Hypophosphatemic nephrolithiasis/osteoporosis 2, (NPHLOP2)	612287	AD	6	28
29	VDR	Vitamin D (1,25-dihydroxyvitamin D3) receptor	NM_000376.2	Idiopathic hypercalciuria	277440	AD	11	29
30	XDH	Xanthine dehydrogenase	NM_000379.3	Xanthinuria, type 1	278300	AR	36	30

AD, autosomal dominant; AR, autosomal recessive.

REFERENCES

1. Purdue PE, Allsop J, Isaya G, Rosenberg LE, Danpure CJ. Mistargeting of peroxisomal L-alanine:glyoxylate aminotransferase to mitochondria in primary hyperoxaluria patients depends upon activation of a cryptic mitochondrial targeting sequence by a point mutation. *Proc Natl Acad Sci U S A*. Dec 1 1991;88(23):10900-10904.
2. Hidaka Y, Palella TD, O'Toole TE, Tarle SA, Kelley WN. Human adenine phosphoribosyltransferase. Identification of allelic mutations at the nucleotide level as a cause of complete deficiency of the enzyme. *J Clin Invest*. Nov 1987;80(5):1409-1415.
3. Smith AN, Skaug J, Choate KA, et al. Mutations in ATP6N1B, encoding a new kidney vacuolar proton pump 116-kD subunit, cause recessive distal renal tubular acidosis with preserved hearing. *Nat Genet*. Sep 2000;26(1):71-75.

4. Karet FE, Finberg KE, Nelson RD, et al. Mutations in the gene encoding B1 subunit of H⁺-ATPase cause renal tubular acidosis with sensorineural deafness. *Nat Genet.* Jan 1999;21(1):84-90.
5. Venta PJ, Welty RJ, Johnson TM, Sly WS, Tashian RE. Carbonic anhydrase II deficiency syndrome in a Belgian family is caused by a point mutation at an invariant histidine residue (107 His----Tyr): complete structure of the normal human CA II gene. *Am J Hum Genet.* Nov 1991;49(5):1082-1090.
6. Pearce SH, Williamson C, Kifor O, et al. A familial syndrome of hypocalcemia with hypercalciuria due to mutations in the calcium-sensing receptor. *N Engl J Med.* Oct 10 1996;335(15):1115-1122.
7. Lloyd SE, Pearce SH, Fisher SE, et al. A common molecular basis for three inherited kidney stone diseases. *Nature.* Feb 01 1996;379(6564):445-449.
8. Simon DB, Bindra RS, Mansfield TA, et al. Mutations in the chloride channel gene, CLCNKB, cause Bartter's syndrome type III. *Nat Genet.* Oct 1997;17(2):171-178.
9. Simon DB, Lu Y, Choate KA, et al. Paracellin-1, a renal tight junction protein required for paracellular Mg²⁺ resorption. *Science.* Jul 2 1999;285(5424):103-106.
10. Konrad M, Schaller A, Seelow D, et al. Mutations in the tight-junction gene claudin 19 (CLDN19) are associated with renal magnesium wasting, renal failure, and severe ocular involvement. *Am J Hum Genet.* Nov 2006;79(5):949-957.
11. Schlingmann KP, Kaufmann M, Weber S, et al. Mutations in CYP24A1 and idiopathic infantile hypercalcemia. *N Engl J Med.* Aug 4 2011;365(5):410-421.
12. Jaureguiberry G, De la Dure-Molla M, Parry D, et al. Nephrocalcinosis (enamel renal syndrome) caused by autosomal recessive FAM20A mutations. *Nephron Physiol.* 2012;122(1-2):1-6.
13. Cramer SD, Ferree PM, Lin K, Milliner DS, Holmes RP. The gene encoding hydroxypyruvate reductase (GRHPR) is mutated in patients with primary hyperoxaluria type II. *Hum Mol Genet.* Oct 1999;8(11):2063-2069.
14. Hamilton AJ, Bingham C, McDonald TJ, et al. The HNF4A R76W mutation causes atypical dominant Fanconi syndrome in addition to a beta cell phenotype. *J Med Genet.* Mar 2014;51(3):165-169.
15. Belostotsky R, Seboun E, Idelson GH, et al. Mutations in DHDPSL are responsible for primary hyperoxaluria type III. *Am J Hum Genet.* Sep 10 2010;87(3):392-399.
16. Davidson BL, Tarle SA, Van Antwerp M, et al. Identification of 17 independent mutations responsible for human hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency. *Am J Hum Genet.* May 1991;48(5):951-958.
17. Simon DB, Karet FE, Rodriguez-Soriano J, et al. Genetic heterogeneity of Bartter's syndrome revealed by mutations in the K⁺ channel, ROMK. *Nat Genet.* Oct 1996;14(2):152-156.
18. Reilly DS, Lewis RA, Ledbetter DH, Nussbaum RL. Tightly linked flanking markers for the Lowe oculocerebrorenal syndrome, with application to carrier assessment. *Am J Hum Genet.* May 1988;42(5):748-755.
19. Simon DB, Karet FE, Hamdan JM, DiPietro A, Sanjad SA, Lifton RP. Bartter's syndrome, hypokalaemic alkalosis with hypercalciuria, is caused by mutations in the Na-K-2Cl cotransporter NKCC2. *Nat Genet.* Jun 1996;13(2):183-188.
20. Enomoto A, Kimura H, Chairoungdua A, et al. Molecular identification of a renal urate anion exchanger that regulates blood urate levels. *Nature.* May 23 2002;417(6887):447-452.
21. Gee HY, Jun I, Braun DA, et al. Mutations in SLC26A1 Cause Nephrolithiasis. *Am J Hum Genet.* Jun 02 2016;98(6):1228-1234.
22. Matsuo H, Chiba T, Nagamori S, et al. Mutations in glucose transporter 9 gene SLC2A9 cause renal hypouricemia. *Am J Hum Genet.* Dec 2008;83(6):744-751.
23. Prie D, Huart V, Bakouh N, et al. Nephrolithiasis and osteoporosis associated with hypophosphatemia caused by mutations in the type 2a sodium-phosphate cotransporter. *N Engl J Med.* Sep 26 2002;347(13):983-991.
24. Lorenz-Depiereux B, Benet-Pages A, Eckstein G, et al. Hereditary hypophosphatemic rickets with hypercalciuria is caused by mutations in the sodium-phosphate cotransporter gene SLC34A3. *Am J Hum Genet.* Feb 2006;78(2):193-201.

25. Calonge MJ, Gasparini P, Chillaron J, et al. Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. *Nat Genet.* Apr 1994;6(4):420-425.
26. Bruce LJ, Cope DL, Jones GK, et al. Familial distal renal tubular acidosis is associated with mutations in the red cell anion exchanger (Band 3, AE1) gene. *J Clin Invest.* Oct 1 1997;100(7):1693-1707.
27. Feliubadalo L, Font M, Purroy J, et al. Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. *Nat Genet.* Sep 1999;23(1):52-57.
28. Karim Z, Gerard B, Bakouh N, et al. NHERF1 mutations and responsiveness of renal parathyroid hormone. *N Engl J Med.* Sep 11 2008;359(11):1128-1135.
29. Scott P, Ouimet D, Valiquette L, et al. Suggestive evidence for a susceptibility gene near the vitamin D receptor locus in idiopathic calcium stone formation. *J Am Soc Nephrol.* May 1999;10(5):1007-1013.
30. Ichida K, Amaya Y, Kamatani N, Nishino T, Hosoya T, Sakai O. Identification of two mutations in human xanthine dehydrogenase gene responsible for classical type I xanthinuria. *J Clin Invest.* May 15 1997;99(10):2391-2397.