

## KidneySeq™ Disorders of Tubular Ion Transport

Cost: \$2000 | CPT Code: 81455

Disease	Inheritance Pattern	Gene(s)
Adrenal hyperplasia, congenital (11b-OH deficiency)	AR	<i>CYP11B1</i>
17α-hydroxylase/17,20-lyase deficiency*	AR	<i>CYP17A1*</i>
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency*	AR	<i>CYP21A1P, CYP21A2*</i>
Apparent mineralocorticoid excess	AR	<i>HSD11B2</i>
APRT deficiency (stones and ESRD)	AR	<i>APRT</i>
Bartter syndrome	AD	<i>CASR</i>
	AR	<i>BSND, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1</i>
Childhood CLD with salt losing tubulopathy		<i>TFCP2L1</i>
Cystinosis	AR	<i>CTNS</i>
Cystinuria	AD, AR	<i>SLC3A1, SLC34A1, SLC7A9</i>
Dent disease	XLR	<i>CLCN5, OCRL</i>
Donnai-Barrow syndrome*	AR	<i>LRP2*</i>
Familial hypocalciuric hypercalcemia	AD	<i>CaSR</i>
Fanconi-Bickel syndrome	AR	<i>SLC2A2</i>
Fanconi syndrome, generalized proximal defect	AD	<i>EHHADH, HNF4A</i>
	AR	<i>ATP7B, CTNS, FAH, GATM, SLC34A1</i>
	XLR	<i>CLCN5</i>
Gitelman syndrome	AR	<i>CLCNKB, SLC12A3</i>
Glucosuria, renal	AD, AR	<i>SLC5A1, SLC5A2</i>
Hypoaldosteronism, congenital	AR	<i>CYP11B2</i>
Hypomagnesemia, renal	AD	<i>CNNM2, FXYP2</i>
	AR	<i>CLDN16, CLDN19, EGF, HNF1B, KCNJ10, TRPM6</i>
Hyperaldosteronism, familial	AD	<i>CACNA1D, CACNA1H, CLCN2, CYP11B1, KCNJ5</i>
Hyperaldosteronism, glucocorticoid remediable	AD	<i>CYP11B1-CYP11B2 fusion</i>
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome (HUPRAS)	AR	<i>SARS2</i>
Hypercalciuria	AD	<i>ADCY10</i>
Hyperoxaluria, primary	AR	<i>AGXT, GRHPR, HOGA1</i>
Hypertension with hyperkalemia (Gordon's syndrome), Pseudohypoaldosteronism II	AD	<i>CUL3, KLHL3, WNK1, WNK4</i>
	AR	<i>KLHL3</i>
Hypocalcemia, autosomal dominant	AD	<i>CASR</i>
Hypokalemic- salt-losing nephropathy	AR	<i>CLDN10, KCNJ10</i>
Hypophosphatemic rickets	AD	<i>FGF23, SGK3*</i>
	AR	<i>DMP1, CYP27B1, ENPP1, SLC34A3, VDR</i>
	XLR	<i>CLCN5</i>
	XLD	<i>PHEX</i>
Hypouricemia, renal	AD	<i>SLC2A9</i>
	AR	<i>SLC22A12</i>
Liddle syndrome (AD) (pseudoprimary hyperaldosteronism)	AD	<i>SCNN1B, SCNN1G</i>
Nephrogenic diabetes insipidus	AD, AR	<i>AQP2</i>
	XLR	<i>AVPR2</i>
Nephrogenic syndrome of inappropriate antidiuresis	XLR	<i>AVPR2</i>
Nephrolithiasis/osteoporosis, hypophosphatemic	AD	<i>SLC9A3R1</i>
Pseudohypoaldosteronism I	AD	<i>NR3C2</i>
	AR	<i>SCNN1A, SCNN1B, SCNN1G</i>
Renal tubular acidosis, distal	AD, AR	<i>ATP6V0A4, ATP6V1B1, ATP6V1C2*, FOXI1, SLC4A1, WDR72*</i>
Renal tubular acidosis, proximal	AD	<i>EHHADH, HNF4A</i>
	AR	<i>ATP7B, CTNS, FAH, SLC34A1, SLC4A4</i>
	XLR	<i>ATP7B, CLCN5</i>
	AR	<i>SCNN1A, SCNN1B, SCNN1G</i>

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Renal tubular acidosis, type IV	AD	<i>CUL3, KLHL3, NR3C2, WNK1, WNK4</i>
Renal tubular acidosis, with osteopetrosis	AR	<i>CA2</i>
Renal tubular disease, hypertension related		<i>NEDD4L</i>
SESAME syndrome / East syndrome	AR	<i>KCNJ10</i>

\*New to KidneySeq™