

The following applies to RSCGP / Nephrocalcinosis, Nephrolithiasis, and Renal Electrolyte Imbalance Gene Panel, Varies. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/-10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis may cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from December 2022 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, or genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ABCC6</i>	NM_001171.5	-	CNV analysis in exons 1–9, 30–31 is not performed
<i>ADCY10</i>	NM_018417.6	-	-
<i>AGXT</i>	NM_000030.3	-	CNV analysis in exon 1 is not performed
<i>ALPL</i>	NM_000478.6	chr1:21896768_21896787del (c.793-30_793-11del)	-
<i>AP2S1</i>	NM_004069.5	-	-
<i>APRT</i>	NM_000485.3	-	-
<i>AQP2</i>	NM_000486.5	-	-
<i>ATP6V0A4</i>	NM_020632.3	-	-
<i>ATP6V1B1</i>	NM_001692.4	-	-
<i>ATP7B</i>	NM_000053.4	chr13:52586924-52585484 covers 5'UTR region	-
<i>AVP</i>	NM_000490.5	-	-
<i>AVPR2</i>	NM_000054.6	-	-
<i>BSND</i>	NM_057176.3	-	-
<i>CA2</i>	NM_000067.3	-	-
<i>CASR</i>	NM_000388.4	-	-
<i>CLCN5</i>	NM_000084.5	-	-
<i>CLCNKA</i>	NM_004070.4	-	-
<i>CLCNKB</i>	NM_000085.5	-	CNV analysis in exon 20 is not performed
<i>CLDN16</i>	NM_006580.3	-	-
<i>CLDN19</i>	NM_148960.3	-	-
<i>CNNM2</i>	NM_017649.5	-	-
<i>CUL3</i>	NM_003590.5	-	-
<i>CYP11B1</i>	NM_000497.3	-	-
<i>CYP11B2</i>	NM_000498.3	-	CNV analysis in exon 9 is not performed
<i>CYP24A1</i>	NM_000782.5	-	CNV analysis in exons 6, 9 is not performed
<i>CYP27B1</i>	NM_000785.4	-	-
<i>CYP2R1</i>	NM_024514.4	-	-
<i>DMP1</i>	NM_004407.4	-	CNV analysis in exon 3 is not performed
<i>EGF</i>	NM_001963.6	-	-
<i>ENPP1</i>	NM_006208.3	-	CNV analysis in exons 10, 22 is not performed
<i>FAM20A</i>	NM_017565.4	-	-
<i>FGF23</i>	NM_020638.3	-	-
<i>FOXI1</i>	NM_012188.5	-	-

**Targeted Genes and Methodology Details
for Nephrocalcinosis, Nephrolithiasis,
and Renal Electrolyte Imbalance Gene Panel** (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FXVD2</i>	NM_001680.5 NM_021603.4	-	-
<i>GALNT3</i>	NM_004482.4	-	-
<i>GATA3</i>	NM_001002295.2	-	-
<i>GNA11</i>	NM_002067.5	-	-
<i>GRHPR</i>	NM_012203.2	-	-
<i>HNF4A</i>	NM_175914.4	-	-
<i>HOGA1</i>	NM_138413.4	-	-
<i>HPRT1</i>	NM_000194.3	chrX:133594415C>T (c.27+47C>T)	CNV analysis in exon 5 is not performed
<i>KCNJ1</i>	NM_000220.5	-	-
<i>KCNJ5</i>	NM_000890.5	-	-
<i>KL</i>	NM_004795.4	-	-
<i>KLHL3</i>	NM_017415.3	-	-
<i>MAGED2</i>	NM_177433.3	-	-
<i>MOCOS</i>	NM_017947.4	-	-
<i>NR3C2</i>	NM_000901.5	-	-
<i>OCRL</i>	NM_000276.4	-	CNV analysis in exons 3, 16 is not performed
<i>PHEX</i>	NM_000444.6	chrX:22266301A>G (c.*231A>G)	CNV analysis in exon 17 is not performed
<i>PRPS1</i>	NM_002764.4	-	-
<i>SCNN1A</i>	NM_001038.6	-	-
<i>SCNN1B</i>	NM_000336.3	-	-
<i>SCNN1G</i>	NM_001039.4	-	-
<i>SLC2A9</i>	NM_020041.3	-	-
<i>SLC3A1</i>	NM_000341.4	-	-
<i>SLC4A1</i>	NM_000342.4	-	-
<i>SLC4A4</i>	NM_001098484.3	-	-
<i>SLC7A9</i>	NM_014270.5	-	-
<i>SLC9A3R1</i>	NM_004252.5	-	-
<i>SLC12A1</i>	NM_000338.3	-	-
<i>SLC12A3</i>	NM_000339.3	chr16: 56917770C>T (c.1670-191C>T)	-
<i>SLC22A12</i>	NM_144585.4	-	-
<i>SLC26A1</i>	NM_213613.4	-	-
<i>SLC34A1</i>	NM_003052.5	-	-
<i>SLC34A3</i>	NM_080877.2	-	-
<i>TRPM6</i>	NM_017662.5	-	-
<i>UMOD</i>	NM_003361.3	-	-
<i>VDR</i>	NM_001017535.1	-	-
<i>WNK1</i>	NM_018979.4	-	-
<i>WNK4</i>	NM_032387.5	-	-
<i>XDH</i>	NM_000379.4	-	-

Effective Date	Version	Synopsis of Test Change
12/22/2022	V2	Added genes <i>AP2S1</i> and <i>FXVD2</i> . Removed genes <i>APOL1</i> and <i>FREM2</i> .