MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Nephrocalcinosis, Nephrolithiasis, and Renal Electrolyte Imbalance Gene Panel

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
ABCC6	NM_001171.5	-	CNV analysis in exons 1–9, 30–31 is not performed
ADCY10	NM_018417.6	-	-
AGXT	NM_000030.3	-	CNV analysis in exon 1 is not performed
ALPL	NM_000478.6	chr1:21896768_21896787del (c.793-30_793-11del)	-
AP2S1	NM_004069.5	-	-
APRT	NM_000485.3	-	-
AQP2	NM_000486.5	-	-
ATP6V0A4	NM_020632.3	-	-
ATP6V1B1	NM_001692.4	-	-
ATP7B	NM_000053.4	chr13:52586924-52585484 covers 5'UTR region	-
AVP	NM_000490.5	-	-
AVPR2	NM_000054.6	-	-
BSND	NM_057176.3	-	-
CA2	NM_000067.3	-	-
CASR	NM_000388.4	-	-
CLCN5	NM_000084.5	-	-
CLCNKA	NM_004070.4	-	-
CLCNKB	NM_000085.5	-	CNV analysis in exon 20 is not performed
CLDN16	NM_006580.3	-	-
CLDN19	NM_148960.3	-	-
CNNM2	NM_017649.5	-	-
CUL3	NM_003590.5	-	-
CYP11B1	NM_000497.3	-	-
CYP11B2	NM_000498.3	-	CNV analysis in exon 9 is not performed
CYP24A1	NM_000782.5	-	CNV analysis in exons 6, 9 is not performed
CYP27B1	NM_000785.4	-	-
CYP2R1	NM_024514.4	-	-
DMP1	NM_004407.4	-	CNV analysis in exon 3 is not performed
EGF	NM_001963.6	-	-
ENPP1	NM_006208.3	-	CNV analysis in exons 10, 22 is not performed
FAM20A	NM_017565.4	-	-
FGF23	NM_020638.3	-	-
FOXI1	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
FXYD2	NM_001680.5 NM_021603.4	-	-
GALNT3	NM_004482.4	-	-
GATA3	NM_001002295.2	-	-
GNA11	NM_002067.5	-	-
GRHPR	NM_012203.2	-	-
HNF4A	NM_175914.4	-	-
HOGA1	NM_138413.4	-	-
HPRT1	NM_000194.3	chrX:133594415C>T (c.27+47C>T)	CNV analysis in exon 5 is not performed
KCNJ1	NM_000220.5	-	-
KCNJ5	NM_000890.5	-	-
KL	NM_004795.4	-	-
KLHL3	NM_017415.3	-	-
MAGED2	NM_177433.3	-	-
MOCOS	NM_017947.4	-	-
NR3C2	NM_000901.5	-	-
OCRL	NM_000276.4	-	CNV analysis in exons 3, 16 is not performed
PHEX	NM_000444.6	chrX:22266301A>G (c.*231A>G)	CNV analysis in exon 17 is not performed
PRPS1	NM_002764.4	-	-
SCNN1A	NM_001038.6	-	-
SCNN1B	NM_000336.3	-	-
SCNN1G	NM_001039.4	-	-
SLC2A9	NM_020041.3	-	-
SLC3A1	NM_000341.4	-	-
SLC4A1	NM_000342.4	-	-
SLC4A4	NM_001098484.3	-	-
SLC7A9	NM_014270.5	-	-
SLC9A3R1	NM_004252.5	-	-
SLC12A1	NM_000338.3	-	-
SLC12A3	NM_000339.3	chr16: 56917770C>T (c.1670-191C>T)	-
SLC22A12	NM_144585.4	-	-
SLC26A1	NM_213613.4	-	-
SLC34A1	NM_003052.5	-	-
SLC34A3	NM_080877.2	-	-
TRPM6	NM_017662.5	-	-
UMOD	NM_003361.3	-	-
VDR	NM_001017535.1	-	-
WNK1	NM_018979.4	-	-
WNK4	NM_032387.5	-	-
XDH	NM_000379.4	-	-

Effective Date	Version	Synopsis of Test Change
12/22/2022	V2	Added genes *AP2S1* and *FXYD2*. Removed genes *APOL1* and *FREM2*.