MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Focal Segmental Glomerulosclerosis and Nephrotic Syndrome Gene Panel

The following applies to RFSGS / Focal Segmental Glomerulosclerosis (FSGS) and Nephrotic Syndrome 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 (CNV) 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 August 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
ACTN4	NM_004924.6	chr:39138352C> T (c.-34C>T)	-
ALG1	NM_019109.5	-	-
ANLN	NM_018685.5	-	-
APOL1	NM_003661.4	*APOL1* gene risk haplotype: G1 and G2 allele	Analyses for sequence variants and CNV is not performed for the gene except for the specified risk variants
ARHGAP24	NM_001025616.3	-	CNV analysis in exon 10 is not performed
ARHGDIA	NM_001185077.3	-	-
CD2AP	NM_012120.3	-	CNV analysis in exon 17 is not performed
CLCN5	NM_000084.5	-	-
COL4A3	NM_000091.5	-	-
COL4A4	NM_000092.5	-	-
COL4A5	NM_000495.5	-	-
COQ2	NM_015697.8	-	-
COQ6	NM_182476.3	-	-
COQ8B	NM_024876.4	-	-
CRB2	NM_173689.7	-	-
CUBN	NM_001081.4	-	-
DGKE	NM_003647.3	chr17: g.54925466A>G (c.888+40A>G)	-
EMP2	NM_001424.6	-	-
FAN1	NM_014967.5	-	-
FAT1	NM_005245.4	-	-
FN1	NM_212482.3	-	-
INF2	NM_022489.4	-	-
ITGA3	NM_002204.4	-	-
ITGB4	NM_000213.5	-	-
KANK2	NM_001136191.3	-	-

Targeted Genes and Methodology Details for Focal Segmental Glomerulosclerosis and Nephrotic Syndrome Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
LAMA5	NM_005560.6	-	-
LAMB2	NM_002292.4	-	-
LMX1B	NM_002316.4	-	-
MAGI2	NM_012301.4	-	CNV analysis in exon 11 is not performed
MYH9	NM_002473.5	-	-
MYO1E	NM_004998.4	-	-
NPHS1	NM_004646.3	-	-
NPHS2	NM_014625.4	-	-
NUP85	NM_024844.5	-	-
NUP93	NM_014669.5	-	-
NUP107	NM_020401.4	-	CNV analysis in exons 18 and 20 is not performed
NUP133	NM_018230.3	chr1: 229577798A>T (c.3335-11T>A)	-
NUP160	NM_015231.2	-	CNV analysis in exon 33 is not performed
NUP205	NM_015135.3	-	-
OCRL	NM_000276.4	-	CNV analysis in exons 3 and 16 is not performed
PAX2	NM_003987.4	-	-
PAX2	NM_003988.5	-	-
PDSS2	NM_020381.4	-	CNV analysis in exon 7 is not performed
PLCE1	NM_016341.4	-	-
PLCG2	NM_002661.5	-	-
PMM2	NM_000303.3	chr16:8891573G>T (c.-167G>T); chr16: .8926102C>T (c.640-15479C>T)	-
PODXL	NM_001018111.3	-	CNV analysis in exon 3 is not performed
PTPRO	NM_030667.3	-	CNV analysis in exon 18 is not performed
SCARB2	NM_005506.4	-	-
SGPL1	NM_003901.4	-	-
SMARCAL1	NM_014140.4	-	-
TBC1D8B	NM_017752.3	-	-
TRPC6	NM_004621.6	-	CNV analysis in exon 10 is not performed
TTC21B	NM_024753.5	-	-
WDR73	NM_032856.4	-	-
WT1	NM_024426.6	-	-
ZMPSTE24	NM_005857.5	-	-