MAYO CLINIC LABORATORIES

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

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. To verify if a specific region/exon/variant is covered by this assay or to confirm transcript version used, contact a laboratory genetic counselor 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 | - | - |
| *ITGA3* | NM_002204.4 | - | - |
| *ITGB4* | NM_000213.5 | - | - |
| *KANK2* | NM_001136191.3 | - | - |
| *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 | - | - |

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| Gene | Reference Transcript | Additional Evaluations | Technical Limitations |
|---|---|---|---|
| *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, 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, 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 | - | - |