MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Alport 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	Technical Limitations
COL4A3	NM_000091.5	-
COL4A4	NM_000092.5	-
COL4A5	NM_000495.5	-
COL4A6	NM_001847.4	CNV analysis in exon 1 is not performed