MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Bartter 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
BSND	NM_057176.3	-
CLCNKA	NM_004070.4	-
CLCNKB	NM_000085.5	CNV analysis in exon 20 is not performed
KCNJ1	NM_000220.5	-
MAGED2	NM_177433.3	-
SLC12A1	NM_000338.3	-