MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Neuronal Ceroid Lipofuscinosis (Batten Disease) Gene Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
ATP13A2	NM_022089.4
CLN3	NM_001042432.1
CLN5	NM_006493.4
CLN6	NM_017882.3
CLN8	NM_018941.4
CTSD	NM_001909.5
CTSF	NM_003793.4
CTSK	NM_000396.4

Gene	Reference Transcript[a]
DNAJC5	NM_025219.3
GRN	NM_002087.3
KCTD7	NM_153033.4
MFSD8	NM_152778.3
PANK2	NM_153638.3
PPT1	NM_000310.3
SGSH	NM_000199.5
TPP1	NM_000391.4

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.