MAYO CLINIC LABORATORIES

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

The following applies to NCLGP / Neuronal Ceroid Lipofuscinosis (Batten Disease) Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 30 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select noncoding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants 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 February 2021 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest to providers as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

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.