

Whole Exome Sequencing

Gene package Ceroidlipofuscinosis (CLN), version 1.6, 25-2-2022



Technical information

DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ATP13A2	610513	100	99.76	97.60	83.45
CLN3	607042	100	98.64	91.76	88.17
CLN5	608102	93.14	93.14	88.00	73.62
CLN6	606725	100	100	100	96.55
CLN8	607837	100	100	100	100
CTSD	116840	100	93.79	93.79	90.47
CTSF	603539	95.77	88.58	86.39	83.35
DNAJC5	611203	100	100	98.81	82.05
GRN	138945	100	100	100	97.18
KCTD7	611725	100	100	100	98.73
MFSD8	611124	100	100	100	97.84
PPT1	600722	100	100	100	96.71
TPP1	607998	100	98.87	96.77	93.28

- OMIM release used: 18-2-2021

- The statistics above are based on a set of 104 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x