Whole Exome Sequencing
Gene package Ceroïdlipofuscinosis (CLN), version 1.4, 26-2-2021

Technical information

DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants are being missed using this technology. 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ATP13A2	610513	100	99.76	96.37	77.46
CLN3	607042	100	97.89	91.51	85.56
CLN5	608102	93.14	93.14	85.77	65.20
CLN6	606725	100	100	100	96.27
CLN8	607837	100	100	100	99.67
CTSD	116840	99.86	93.79	93.64	88.98
CTSF	603539	94.74	87.79	86.39	82.89
DNAJC5	611203	100	100	100	81.90
GRN	138945	100	100	100	97.13
KCTD7	611725	100	100	100	98.20
MFSD8	611124	100	100	98.86	96.20
PPT1	600722	100	100	100	95.20
TPP1	607998	100	99.44	97.38	93.18

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x