Whole Exome Sequencing
Gene package Ceroïdlipofuscinosis (CLN), version 1.1, 22-11-2017

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations 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	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x	% covered >30x
ATP13A2	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693	610513	NM_022089.2	79	100	100	98
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	NM_001042432.1	76	100	100	94
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	NM_006493.2	57	100	100	98
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	NM_017882.2	78	100	98	88
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	NM_018941.3	81	100	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	NM_001909.4	79	100	100	100
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	NM_003793.3	70	100	98	93
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	NM_025219.2	129	100	100	97
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	138945	NM_002087.2	101	100	100	100
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	NM_153033.4	102	100	100	99
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	NM_152778.2	50	100	98	81
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	NM_000310.3	51	100	100	87

TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	NM_000391.3	70	100	100	99

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 96 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x