Whole Exome Sequencing
Gene package Ceroïdlipofuscinosis (CLN), version 1, 17-8-2015

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x
ATP13A2	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693	610513	NM_022089.2	67	100	99
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	NM_001042432.1	68	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	NM_006493.2	73	100	94
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	NM_017882.2	72	100	91
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	NM_018941.3	100	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	NM_001909.4	83	100	96
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	NM_003793.3	43	82	70
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	NM_025219.2	111	100	100
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	138945	NM_002087.2	95	100	100
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	NM_153033.4	90	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	NM_152778.2	75	100	97
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	NM_000310.3	81	100	100
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270	607998	NM_000391.3	79	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	Transcript	median depth	% covered >10x	% covered >20x

- 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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x