Whole Exome Sequencing
Gene package Familial Hemophagocytic Lymphohistiocytosis (fHLH)
Version 3.1, 25-2-2022

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). Additionally, MPLA analysis was performed for fHLH related genes (probemix P028, version B1; MRC Holland. 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
AP3B1	603401	99.05	96.39	94.98	89.23
AP3D1	607246	97.71	97.64	95.96	89.40
CD27	186711	100	100	100	98.11
CD70	602840	100	100	100	94.96
CORO1A	605000	92.29	91.79	90.59	86.61
CTPS1	123860	100	99.91	98.44	94.08
FAAP24	610884	100	100	100	97.52
ITK	186973	100	100	100	98.77
LYST	606897	100	99.99	99.68	96.53
MAGT1	300715	100	98.65	94.16	90.58
PRF1	170280	100	100	100	100
RAB27A	603868	100	100	100	100
RASGRP1	603962	100	100	100	99.95
RC3H1	609424	100	99.46	98.51	93.85
RHOG	179505	100	100	100	100
SH2D1A	300490	100	100	100	100
STX11	605014	100	100	100	100
STXBP2	601717	100	97.40	91.65	77.53
UNC13D	608897	100	100	99.12	91.74
XIAP	300079	100	100	100	93.11

- OMIM release used: 23-9-2021
- 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 (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x