Whole Exome Sequencing
Gene package Familial Hemophagocytic Lymphohistiocytosis (fHLH)
Version 1, 31-1-2020

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/). 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. Additionally, MPLA analysis was performed for fHLH related genes (probemix P028, version B1; MRC Holland). 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	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AP3B1	603401	66	100	98	87
AP3D1	607246	103	99	98	97
CD27	186711	95	100	100	100
CD70	602840	100	100	100	100
CORO1A	605000	122	92	92	91
CTPS1	123860	70	100	100	97
FAAP24	610884	97	100	100	100
ITK	186973	76	100	100	95
LYST	606897	72	100	99	95
MAGT1	300715	47	100	99	85
PRF1	170280	120	100	100	100
RAB27A	603868	54	100	100	91
RASGRP1	603962	70	100	100	98
SH2D1A	300490	56	100	98	83
STX11	605014	308	100	100	100
STXBP2	601717	121	100	100	100
UNC13D	608897	100	100	100	100
XIAP	300079	47	100	95	80

- Gene symbols according HGCN
- OMIM release used: 8-9-2019

- The statistics above are based on a set of 100 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 , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x