Whole Exome Sequencing
Gene package Familial Hemophagocytic Lymphohistiocytosis (fHLH)
Version 1.1, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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 Alissa Interpret 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AP3B1	603401	58	93	79	66
AP3D1	607246	241	99	98	97
CD27	186711	205	100	100	100
CD70	602840	165	100	100	99
CORO1A	605000	287	92	92	92
CTPS1	123860	96	100	100	99
FAAP24	610884	100	100	100	100
ITK	186973	104	100	100	96
LYST	606897	87	97	92	89
MAGT1	300715	69	93	87	78
PRF1	170280	383	100	100	100
RAB27A	603868	88	100	100	97
RASGRP1	603962	138	100	100	100
SH2D1A	300490	81	100	95	85
STX11	605014	478	100	100	100
STXBP2	601717	220	100	100	100
UNC13D	608897	248	100	100	100
XIAP	300079	63	94	89	81

- 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

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