Whole Exome Sequencing
Gene package Very early onset inflammatory bowel disease / congenital diarrhea, version 5.2, 25-2-2022

Technical information
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). 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	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ACTG2	102545	100	100	100	100
ADAM17	603639	100	99.70	98.07	91.74
AIRE	607358	100	99.53	96.71	88.36
ALPI	171740	100	100	100	97.87
ANKZF1	617541	100	99.18	95.73	86.32
ARPC1B	604223	100	100	100	99.04
ARX	300382	84.88	79.12	68.56	50.95
BACH2	605394	100	100	100	99.02
CARD8	609051	100	100	100	99.13
CARMIL2	610859	97.18	94.69	93.13	86.05
CD19	107265	100	99.95	98.05	92.06
CD40	109535	100	100	100	100
CD40LG	300386	100	100	100	100
CD55	125240	100	97.01	94.12	84.45
CLMP	611693	100	100	100	96.19
COG6	606977	100	100	100	97.68
CR2	120650	100	100	100	98.43
CTLA4	123890	100	100	100	100
CYBA	608508	95.74	84.68	71.77	50.92
CYBB	300481	100	100	99.95	95.66
CYBC1	No ID	100	99.71	93.61	84.29
DEF6	610094	100	98.20	94.82	89.23
DGAT1	604900	95.12	88.94	87.80	87.80
DOCK2	603122	100	99.82	99.04	97.48

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DOCK8	611432	100	99.94	99.37	96.42
DUOX2	606759	100	97.73	96.13	90.83
EGFR	131550	100	100	98.97	96.26
ELF4	300775	100	99.46	96.84	87.62
EPCAM	185535	100	100	98.35	91.44
FOXP3	300292	100	100	98.15	78.49
GPIHBP1	612757	100	100	100	96.44
GUCY2C	601330	100	100	99.68	97.69
HPS1	604982	100	100	99.68	90.74
HPS4	606682	100	100	100	96.66
HPS6	607522	100	100	98.59	84.20
ICOS	604558	100	100	100	100
IKBKG	300248	36.54	25.95	25.35	20.68
IL10	124092	100	100	100	100
IL10RA	146933	100	100	98.77	94.77
IL10RB	123889	100	95.38	93.54	90.04
IL21	605384	100	100	98.98	88.82
IL21R	605383	100	100	100	97.58
IL2RA	147730	100	100	100	100
IL2RB	146710	100	100	98.85	93.62
ITCH	606409	98.24	96.53	94.90	91.64
ITGB2	600065	100	98.31	95.32	83.70
LCT	603202	100	100	99.86	98.74
LRBA	606453	100	99.46	99.40	97.12
MALT1	604860	95.96	92.09	90.24	89.69
MEFV	608107	100	100	99.57	94.81
MPI	154550	100	100	100	100
MVK	251170	100	100	100	96.15
MYO5B	606540	100	100	99.47	95.36
NCF1	608512	58.99	50.72	35.79	21.19
NCF2	608515	100	100	100	98.03
NCF4	601488	100	100	99.22	84.51
NEUROG3	604882	100	100	97.51	91.99
NFKBIA	164008	100	100	100	95.52
NLRC4	606831	100	100	100	100
NOD2	605956	100	100	99.81	96.90
OTULIN	615712	91.60	85.62	85.62	82.65
PCSK1	162150	100	100	100	97.83
PLA2G4A	600522	100	100	100	98.36
PLCG2	600220	100	100	100	96.79
PLVAP	607647	100	100	98.13	89.35
PNLIP	246600	100	100	100	98.26
POLA1	312040	100	100	99.02	92.76
PTPN2	176887	100	93.86	93.86	92.66

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RAC2	602049	100	100	100	98.85
RFX6	612659	100	100	100	98.48
RIPK1	603453	100	100	100	95.68
SAR1B	607690	100	96.36	87.96	87.96
SH2D1A	300490	100	100	100	100
SI	609845	100	99.60	97.21	92.96
SKIV2L	600478	100	100	99.13	93.93
SLC10A2	601295	100	100	100	98.54
SLC26A3	126650	100	100	100	99.74
SLC2A2	138160	100	100	100	99.33
SLC37A4	602671	99.87	99.87	99.42	96.62
SLC39A4	607059	100	100	100	100
SLC5A1	182380	100	100	100	96.95
SLC7A7	603593	100	100	100	99.01
SLC9A3	182307	100	98.79	96.66	88.37
SPINT2	605124	100	100	96.65	85.66
STAT1	600555	99.29	97.09	97.09	96.54
STAT3	102582	100	100	100	95.99
STAT5B	604260	100	99.03	96.66	89.84
STX3	600876	100	100	98.13	95.31
STXBP2	601717	100	97.40	91.65	77.53
TCN2	613441	100	100	100	95.04
TGFB1	190180	100	100	100	99.69
TMPRSS15	606635	98.78	92.07	88.28	87.21
TNFAIP3	191163	100	100	99.80	98.30
TNFRSF13B	604907	100	100	98.88	90.14
TNFRSF13C	606269	91.42	67.57	59.97	51.47
TOM1	604700	100	100	100	98.26
TRIM22	606559	100	100	99.60	93.15
TTC37	614589	100	100	100	98.95
TTC7A	609332	100	100	99.67	92.91
TYMP	131222	100	100	97.96	83.49
WAS	300392	100	99.86	94.70	77.23
WIPF1	602357	100	98.61	97.34	83.73
WNT2B	601968	100	100	100	97.34
XIAP	300079	100	100	100	93.11
ZAP70	176947	100	100	98.52	93.32

- OMIM release used: 18-2-2021
- 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

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