Whole Exome Sequencing
Gene package Early-onset enteropathy, version 1, 1-2-2018

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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. 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	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ADAM17	?Inflammatory skin and bowel disease, neonatal, 1, 614328	603639	51	100	96	77
APOB	Hypercholesterolemia, due to ligand-defective apo B, 144010 Hypobetalipoproteinemia, 615558	107730	115	100	100	99
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	42	89	80	65
CD19	Immunodeficiency, common variable, 3, 613493	107265	73	100	100	100
CD40	Immunodeficiency with hyper-IgM, type 3, 606843	109535	73	100	100	100
CD40LG	Immunodeficiency, with hyper-IgM, 308230	300386	55	100	100	97
CTLA4	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700	123890	65	100	100	98
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690	608508	60	99	89	79

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CYBB	Chronic granulomatous disease, 306400 Immunodeficiency 34, mycobacteriosis, 300645	300481	39	100	95	79
DGAT1	?Diarrhea 7, 615863	604900	87	97	93	88
DOCK2	Immunodeficiency 40, 616433	603122	51	100	98	88
DOCK8	Hyper-IgE recurrent infection syndrome, 243700	611432	49	100	94	81
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217	185535	51	100	99	87
FOXP3	{Diabetes mellitus, type I, susceptibility to}, 222100 Immunodysregulation, polyendocrinopathy, and enteropathy, 304790	300292	59	100	100	91
GUCY2C	Diarrhea 6, 614616 Meconium ileus, 614665	601330	55	100	99	84
ICOS	Immunodeficiency, common variable, 1, 607594	604558	42	100	100	88
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640	300248	62	100	100	92
IL10	{Graft-versus-host disease, protection against}, 614395 {HIV-1, susceptibility to}, 609423 {Rheumatoid arthritis, progression of}, 180300	124092	81	100	98	86
IL10RA	Inflammatory bowel disease 28, early onset, 613148	146933	73	100	100	96
IL10RB	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, 612567	123889	42	100	98	81
IL21	?Immunodeficiency, common variable, 11, 615767	605384	52	100	98	80
IL21R	[IgE, elevated level of], 147050 Immunodeficiency, primary, IL21R-related, 615207	605383	78	100	100	95
IL2RA	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367	147730	67	100	100	94
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385	606409	43	96	93	76
ITGB2	Leukocyte adhesion deficiency, 116920	600065	77	100	100	99
LCT	Lactase deficiency, congenital, 223000	603202	84	100	100	98
LRBA	Immunodeficiency, common variable, 8, with autoimmunity, 614700	606453	47	100	97	82
MALT1	Immunodeficiency 12, 615468	604860	51	99	93	77
MEFV	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100	608107	78	100	100	99
MGAM	No OMIM phenotype	154360	62	100	97	89

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MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	49	100	99	89
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	58	100	100	91
MYO5B	Microvillus inclusion disease, 251850	606540	68	100	99	90
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700	608512	98	100	100	98
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710	608515	63	100	100	90
NCF4	?Granulomatous disease, chronic, cytochrome b-positive, type III, 613960	601488	67	100	100	99
NEUROG3	Diarrhea 4, malabsorptive, congenital, 610370	604882	121	100	100	100
NLRC4	Autoinflammation with infantile enterocolitis, 616050 ?Familial cold autoinflammatory syndrome 4, 616115	606831	61	100	99	91
NOD2	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321	605956	73	100	99	94
PCSK1	Obesity with impaired prohormone processing, 600955 {Obesity, susceptibility to, BMIQ12}, 612362	162150	48	100	97	80
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468	600220	70	100	99	92
PNLIP	?Pancreatic lipase deficiency, 614338	246600	42	100	98	80
PRSS1	Pancreatitis, hereditary, 167800 Trypsinogen deficiency, 614044	276000	142	100	100	100
RFX6	Mitchell-Riley syndrome, 615710	612659	62	100	97	80
SAR1B	Chylomicron retention disease, 246700	607690	53	100	100	82
SI	Sucrase-isomaltase deficiency, congenital, 222900	609845	45	100	97	80
SKIV2L	Trichohepatoenteric syndrome 2, 614602	600478	87	100	100	100
SLC10A2	Bile acid malabsorption, primary, 613291	601295	61	100	98	85
SLC26A3	Diarrhea 1, secretory chloride, congenital, 214700	126650	52	100	99	88
SLC2A2	{Diabetes mellitus, noninsulin-dependent}, 125853 Fanconi-Bickel syndrome, 227810	138160	47	100	99	86
SLC2A5	No OMIM phenotype	138230	65	100	100	96
SLC37A4	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240	602671	69	100	100	97
SLC39A4	Acrodermatitis enteropathica, 201100	607059	71	100	100	100
SLC5A1	Glucose/galactose malabsorption, 606824	182380	70	100	96	86
SLC7A7	Lysinuric protein intolerance, 222700	603593	54	100	97	88
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420	605124	59	100	100	99

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STAT1	Immunodeficiency 31A, mycobacteriosis, 614892 Immunodeficiency 31B, mycobacterial and viral infections, 613796 Immunodeficiency 31C, 614162	600555	43	100	96	80
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060	102582	60	100	100	91
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578	604260	71	100	99	89
STX3	No OMIM phenotype	600876	53	100	99	87
TMPRSS15	Enterokinase deficiency, 226200	606635	48	100	97	82
TNFRSF13B	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529	604907	55	100	100	96
TNFRSF13C	Immunodeficiency, common variable, 4, 613494	606269	53	100	84	65
TNFSF13	No OMIM phenotype	604472	51	100	100	86
TNFSF13B	No OMIM phenotype	603969	39	100	94	82
TTC37	Trichohepatoenteric syndrome 1, 222470	614589	43	100	97	80
TTC7A	Gastrointestinal defects and immunodeficiency syndrome, 243150	609332	68	100	99	94
WAS	Neutropenia, severe congenital, 300299 Thrombocytopenia, 313900 Thrombocytopenia, intermittent, 313900 Wiskott-Aldrich syndrome, 301000	300392	54	96	84	75
XIAP	Lymphoproliferative syndrome, 2, 300635	300079	52	100	96	85

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 96 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x