

Whole Exome Sequencing

Gene package Metabolic disorders, version 10, 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
AASS	605113	100	100	99.94	98.67
ABAT	137150	93.95	93.95	92.80	82.33
ABCA1	600046	100	100	99.98	99.21
ABCB11	603201	100	100	100	98.67
ABCB4	171060	100	100	100	98.08
ABCB7	300135	100	100	99.98	97.44
ABCD1	300371	100	95.42	90.51	80.47
ABCD4	603214	100	100	99.95	94.88
ABCG2	603756	100	100	100	100
ABCG5	605459	100	99.80	97.99	90.67
ABCG8	605460	100	99.17	96.40	88.00
ABHD12	613599	100	96.57	91.81	82.57
ABHD5	604780	100	100	92.98	91.61
ACACA	200350	100	99.98	99.67	98.00
ACAD8	604773	100	100	100	93.11
ACAD9	611103	100	100	100	99.64
ACADM	607008	100	100	100	93.69
ACADS	606885	100	100	99.30	94.81
ACADSB	600301	99.93	99.87	95.78	93.80
ACADVL	609575	100	99.71	98.00	90.48
ACAT1	607809	100	99.97	96.50	89.04
ACAT2	100678	100	100	100	100
ACBD5	616618	100	99.32	97.67	95.81

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ACO2	100850	100	100	100	100
ACOX1	609751	100	100	100	98.47
ACSF3	614245	100	100	100	98.74
ACY1	104620	100	100	99.67	89.10
ADA	608958	100	100	99.94	92.54
ADCY5	600293	99.14	97.89	96.92	92.30
ADK	102750	100	93.49	93.49	88.94
ADSL	608222	98.31	98.31	98.31	98.31
AGA	613228	100	100	100	97.41
AGK	610345	100	100	100	100
AGL	610860	100	100	99.83	97.29
AGPAT2	603100	100	100	100	93.29
AGPS	603051	99.54	95.91	95.16	92.04
AGXT	604285	100	100	100	92.87
AGXT2	612471	100	100	100	99.51
AHCY	180960	100	100	100	100
AIFM1	300169	100	100	99.17	95.63
AK1	103000	100	100	97.20	80.80
AK2	103020	100	98.15	95.34	89.19
AKR1D1	604741	100	100	99.44	95.03
ALAD	125270	100	100	99.38	95.10
ALAS2	301300	100	100	99.75	94.90
ALDH18A1	138250	100	100	100	98.06
ALDH1A3	600463	100	99.78	95.41	85.77
ALDH3A2	609523	100	100	99.88	96.84
ALDH4A1	606811	100	99.45	97.89	93.74
ALDH5A1	610045	100	99.36	94.61	80.34
ALDH6A1	603178	100	100	100	100
ALDH7A1	107323	100	100	100	99.34
ALDOA	103850	100	100	100	97.00
ALDOB	612724	100	100	100	100
ALG1	605907	100	98.64	95.61	86.08
ALG11	613666	100	100	100	99.65
ALG12	607144	100	100	100	100
ALG13	300776	100	100	100	97.07
ALG14	612866	100	100	100	100
ALG2	607905	100	100	100	97.90
ALG3	608750	100	100	98.86	94.65
ALG6	604566	100	97.83	95.78	95.06
ALG8	608103	100	98.61	92.79	88.19
ALG9	606941	99.91	99.91	98.98	91.73
ALOX12B	603741	100	100	100	98.59
ALPL	171760	100	100	99.83	98.30
AMACR	604489	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
AMN	605799	100	95.84	87.15	69.07
AMPD1	102770	100	100	100	98.92
AMPD2	102771	100	99.89	97.72	87.37
AMT	238310	100	100	100	97.08
APOA1	107680	100	100	100	99.30
APOB	107730	100	99.70	99.35	99.29
APOC2	608083	100	100	100	100
APOE	107741	100	96.53	88.32	61.32
APRT	102600	100	100	100	90.55
APTX	606350	100	100	100	98.02
ARG1	608313	100	100	100	99.70
ARSA	607574	100	100	100	100
ARSB	611542	100	100	99.12	89.26
ASAH1	613468	100	100	99.41	95.80
ASL	608310	100	100	100	96.47
ASNS	108370	100	100	100	98.34
ASPA	608034	100	100	100	100
ASS1	603470	100	100	100	89.02
ATIC	601731	100	100	100	95.94
ATP5F1E	606153	100	100	100	100
ATP6AP1	300197	100	100	93.66	83.51
ATP6VOA2	611716	100	100	99.49	96.93
ATP7A	300011	100	100	99.70	97.72
ATP7B	606882	100	100	99.51	96.63
ATP8B1	602397	100	100	99.79	97.71
ATPAF2	608918	100	100	100	99.22
AUH	600529	100	100	100	100
B3GALNT1	603094	100	100	98.93	88.37
B3GALNT2	610194	100	100	100	96.03
B3GALT6	615291	77.76	74.28	72.00	69.31
B3GAT3	606374	100	100	100	94.34
B3GLCT	610308	100	99.36	94.98	94.20
B4GALNT1	601873	100	97.64	95.30	88.27
B4GALT1	137060	100	100	98.78	81.22
B4GALT7	604327	100	93.64	93.32	84.20
BAAT	602938	100	100	98.86	95.85
BCKDHA	608348	100	98.88	95.74	89.24
BCKDHB	248611	100	100	100	100
BCKDK	614901	100	100	98.83	93.41
BCO1	605748	100	100	100	98.71
BCS1L	603647	100	100	100	100
BHMT	602888	100	99.67	97.53	91.26
BLVRA	109750	100	100	100	100
BMP2	112261	100	99.71	99.43	98.53

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
BOLA3	613183	100	100	100	81.55
BPGM	613896	100	100	100	100
BPNT2	614010	100	100	100	98.64
BTD	609019	100	100	100	100
C1GALT1C1	300611	100	100	100	100
CA5A	114761	100	100	99.81	94.36
CAD	114010	100	100	99.90	96.49
CANT1	613165	100	100	100	98.50
CAT	115500	100	100	100	100
CBLIF	609342	100	100	100	95.88
CBS	613381	100	100	100	100
CCDC115	613734	100	100	98.67	88.28
CD320	606475	100	100	98.78	79.23
CEL	114840	94.41	91.49	87.72	75.66
CERKL	608381	100	100	100	98.82
CERS3	615276	100	100	100	99.22
CETP	118470	100	99.69	97.19	85.44
CHKB	612395	100	100	100	98.26
CHST14	608429	99.35	96.21	94.08	90.85
CHST3	603799	100	100	100	100
CHST6	605294	100	100	100	100
CHSY1	608183	94.20	93.39	92.74	90.55
CIC	612082	100	100	98.96	90.31
CLDN16	603959	100	100	100	100
CLDN19	610036	100	100	100	100
CLN3	607042	100	98.64	91.76	88.17
CLN5	608102	93.14	93.14	88.00	73.62
CLN6	606725	100	100	100	96.55
CLN8	607837	100	100	100	100
CLPB	616254	100	100	100	98.48
CLPS	120105	100	100	100	92.93
CLPX	615611	100	100	100	99.63
CNDP1	609064	100	100	98.35	90.55
CNNM2	607803	100	100	100	98.11
COG1	606973	100	100	100	97.42
COG2	606974	100	100	99.55	91.34
COG4	606976	100	100	100	98.14
COG5	606821	100	98.39	95.95	92.69
COG6	606977	100	100	100	97.68
COG7	606978	100	100	100	98.34
COG8	606979	92.58	92.32	90.49	87.63
COQ2	609825	100	100	99.68	89.02
COQ8A	606980	100	100	100	97.43
COX10	602125	100	100	100	98.57

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
COX15	603646	100	100	98.03	93.23
COX6B1	124089	100	100	100	96.23
CP	117700	99.05	99.05	99.05	98.77
CPOX	612732	100	100	97.47	80.29
CPS1	608307	99.28	99.28	99.28	99.15
CPT1A	600528	100	100	100	98.32
CPT2	600650	100	100	99.23	96.96
CRPPA	614631	100	99.36	96.94	90.76
CRTC1	607536	100	98.29	95.18	78.47
CTH	607657	100	100	99.52	96.36
CTNS	606272	100	93.85	80.32	69.81
CTSA	613111	100	99.85	99.59	92.57
CTSC	602365	100	100	100	100
CTSD	116840	100	93.79	93.79	90.47
CTSK	601105	100	100	100	98.94
CUBN	602997	100	100	99.93	98.53
CYB5R3	613213	100	100	97.43	82.60
CYP27A1	606530	100	100	100	100
CYP27B1	609506	100	100	100	94.45
CYP7A1	118455	100	100	100	100
CYP7B1	603711	100	99.88	93.62	91.33
D2HGDH	609186	100	100	98.48	95.27
DARS2	610956	100	100	100	97.36
DBH	609312	100	99.45	98.66	94.17
DBT	248610	100	99.94	99.88	95.65
DCXR	608347	100	100	100	91.65
DDC	107930	100	98.82	95.38	82.24
DDHD1	614603	99.93	98.53	96.74	94.76
DDOST	602202	100	100	100	98.99
DGAT1	604900	95.12	88.94	87.80	87.80
DGKE	601440	100	100	99.53	94.64
DGUOK	601465	100	100	100	96.86
DHCR24	606418	100	100	100	97.77
DHCR7	602858	100	100	99.42	94.57
DHDDS	608172	100	100	100	93.45
DHFR	126060	100	100	99.49	90.23
DHODH	126064	100	100	100	94.91
DHTKD1	614984	100	100	99.81	97.11
DLAT	608770	100	99.98	98.91	96.09
DLD	238331	100	100	100	98.89
DLST	126063	100	100	100	95.22
DMGDH	605849	100	100	99.74	96.68
DNAJC12	606060	100	100	100	98.70
DNAJC19	608977	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
DNAJC5	611203	100	100	98.81	82.05
DNM1L	603850	100	100	99.23	93.91
DNMT1	126375	100	99.85	98.71	94.39
DOLK	610746	100	100	100	100
DPAGT1	191350	100	100	100	97.76
DPEP1	179780	100	100	99.76	95.05
DPM1	603503	100	100	100	99.38
DPM2	603564	100	100	100	89.16
DPM3	605951	100	100	100	94.95
DPYD	612779	100	100	100	100
DPYS	613326	100	100	99.77	97.73
EBP	300205	100	100	99.55	91.56
ECHS1	602292	100	100	95.05	89.51
EGF	131530	100	100	99.73	96.57
ELOVL4	605512	100	99.81	98.78	92.80
ENO3	131370	97.13	97.13	97.13	95.89
EOGT	614789	100	100	99.95	98.14
EPG5	615068	100	99.90	99.27	95.85
EPHX1	132810	100	100	100	96.05
EPHX2	132811	100	100	98.26	93.94
EPM2A	607566	85.89	81.07	79.79	76.89
ETFA	608053	100	100	100	99.80
ETFB	130410	100	100	95.71	70.06
ETFDH	231675	100	100	97.44	96.76
ETHE1	608451	100	95.84	88.80	87.47
EXT1	608177	100	100	100	99.43
EXT2	608210	100	100	100	99.64
FA2H	611026	100	96.58	89.53	72.53
FAH	613871	100	100	100	99.61
FASTKD2	612322	100	100	100	100
FBP1	611570	100	100	100	100
FECH	612386	100	100	99.87	96.95
FGFR2	176943	100	100	99.68	98.15
FH	136850	100	97.54	94.62	91.21
FKRP	606596	100	100	100	98.50
FKTN	607440	100	100	99.84	97.44
FLAD1	610595	100	100	100	100
FMO3	136132	100	100	100	100
FOLR1	136430	100	100	100	100
FOXRED1	613622	100	100	100	99.40
FTCD	606806	98.36	94.97	92.56	91.02
FUCA1	612280	100	100	100	99.42
FUT2	182100	100	100	100	100
FXD2	601814	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
G6PC1	613742	-	-	-	-
G6PC3	611045	100	100	100	100
G6PD	305900	100	100	98.26	92.43
GAA	606800	100	100	99.68	93.73
GAD1	605363	100	100	100	100
GALC	606890	99.96	99.96	99.96	97.95
GALE	606953	100	100	99.60	93.77
GALK1	604313	100	100	100	96.78
GALNS	612222	100	100	99.41	87.98
GALNT3	601756	100	100	100	99.74
GALT	606999	100	100	100	97.01
GAMT	601240	100	96.09	87.27	77.84
GANAB	104160	100	100	100	98.37
GART	138440	100	100	100	98.29
GATM	602360	100	100	100	99.38
GBA	606463	100	100	100	99.29
GBA2	609471	100	100	99.49	98.09
GBE1	607839	100	100	100	99.38
GCDH	608801	100	100	100	96.94
GCH1	600225	94.02	94.02	94.02	92.99
GCK	138079	100	100	100	87.83
GCLC	606857	100	100	100	99.01
GCSH	238330	100	79.89	72.86	70.19
GFM1	606639	100	100	100	96.47
GFPT1	138292	100	100	100	99.64
GGT1	612346	93.32	79.40	67.72	53.72
GK	300474	100	97.69	90.34	79.21
GLA	300644	100	100	100	98.28
GLB1	611458	100	100	99.74	96.88
GLDC	238300	100	100	99.76	97.44
GLRA1	138491	100	100	100	97.81
GLRX5	609588	100	80.53	67.81	51.76
GLUD1	138130	100	100	100	97.67
GLUL	138290	100	100	100	99.27
GLYCTK	610516	100	100	100	97.11
GM2A	613109	100	100	100	100
GMPPA	615495	100	100	99.67	91.73
GMPPB	615320	100	100	100	99.47
GNE	603824	100	100	100	100
GNMT	606628	100	100	99.79	98.31
GNPAT	602744	100	100	100	96.02
GNPTAB	607840	100	100	99.79	98.44
GNPTG	607838	100	97.58	94.41	93.66
GNS	607664	100	97.83	96.15	93.60

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
GPD1	138420	100	100	100	96.64
GPHN	603930	97.10	97.10	97.04	92.87
GPI	172400	100	100	100	94.52
GPX1	138320	100	100	100	99.54
GRHPR	604296	100	100	98.02	87.03
GSS	601002	100	100	98.29	91.58
GSTZ1	603758	100	100	100	98.92
GUSB	611499	100	100	100	98.65
GYG1	603942	100	100	100	98.80
GYS1	138570	100	100	99.64	96.74
GYS2	138571	100	100	98.39	97.04
HADH	601609	100	100	100	100
HADHA	600890	100	100	100	96.76
HADHB	143450	99.71	98.14	96.86	95.33
HAL	609457	100	100	100	98.36
HAMP	606464	100	100	91.67	54.01
HCFC1	300019	100	100	99.50	95.18
HEXA	606869	100	100	100	98.79
HEXB	606873	100	97.84	95.71	90.79
HFE	613609	100	100	100	96.42
HGD	607474	100	100	100	100
HGSNAT	610453	94.18	93.42	91.23	83.05
HIBCH	610690	100	100	100	94.51
HK1	142600	100	99.94	98.93	93.60
HLCS	609018	86.97	86.97	86.97	86.62
HMBS	609806	100	100	100	97.40
HMGCL	613898	100	100	100	100
HMGCS2	600234	100	99.88	98.83	96.13
HMOX1	141250	100	100	98.96	90.72
HOGA1	613597	100	100	100	91.85
HPD	609695	100	100	100	98.05
HPRT1	308000	100	95.44	93.17	84.11
HPS1	604982	100	100	99.68	90.74
HS6ST1	604846	100	100	100	97.53
HSD11B1	600713	100	100	100	94.28
HSD11B2	614232	88.33	78.33	77.87	75.59
HSD17B10	300256	100	100	100	96.57
HSD17B4	601860	100	98.63	96.80	91.72
HSD3B2	613890	100	100	100	100
HSD3B7	607764	100	100	100	100
HYAL1	607071	100	100	98.17	84.86
IBA57	615316	100	100	100	100
IDH2	147650	100	98.22	91.50	90.70
IDH3B	604526	100	100	100	98.45

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
IDS	300823	100	100	98.17	89.78
IDUA	252800	99.80	90.46	84.30	72.67
IMPDH1	146690	100	98.62	92.40	89.49
INPPL1	600829	98.63	95.71	94.66	87.53
ISCU	611911	100	100	100	97.75
ITPA	147520	100	100	100	99.46
IVD	607036	100	100	100	97.58
KHK	614058	100	100	100	96.16
KYNU	605197	100	100	100	99.70
L2HGDH	609584	100	100	99.62	91.41
LAMP2	309060	100	100	100	91.30
LARGE1	603590	100	100	99.18	96.64
LBR	600024	100	100	100	94.80
LCAT	606967	100	100	98.65	87.92
LCT	603202	100	100	99.86	98.74
LDHA	150000	100	100	100	100
LDLR	606945	100	100	100	98.61
LFNG	602576	100	100	100	90.22
LIPA	613497	100	100	100	100
LIPC	151670	100	100	100	97.91
LIPI	609252	100	100	99.56	90.89
LMBRD1	612625	100	97.14	93.56	87.45
LPIN1	605518	100	99.72	98.87	95.93
LPIN2	605519	100	100	100	98.57
LPL	609708	100	100	100	98.69
LRP2	600073	100	99.96	99.64	97.62
LRPPRC	607544	100	99.78	98.96	96.81
LTC4S	246530	100	85.64	71.45	48.45
LYST	606897	100	99.99	99.68	96.53
MAN1B1	604346	100	100	100	98.20
MAN2B1	609458	100	99.96	97.78	88.49
MANBA	609489	100	100	99.97	99.23
MAOA	309850	100	100	100	97.95
MAT1A	610550	100	100	100	100
MAT2A	601468	100	100	100	100
MCCC1	609010	100	100	100	96.81
MCCC2	609014	100	100	98.84	90.86
MCEE	608419	100	100	100	100
MCOLN1	605248	100	99.78	97.55	90.27
MFSD8	611124	100	100	100	97.84
MGAT2	602616	100	100	100	100
MLYCD	606761	99.27	93.67	88.41	73.50
MMAA	607481	100	100	100	97.23
MMAB	607568	100	100	100	97.20

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
MMACHC	609831	100	100	100	95.30
MMADHC	611935	100	100	100	95.67
MMUT	609058	100	100	100	100
MOCOS	613274	100	100	99.21	94.10
MOCS1	603707	100	100	97.49	93.02
MOCS2	603708	100	100	100	100
MOCS3	609277	100	100	100	100
MOGS	601336	100	100	100	98.42
MPDU1	604041	100	100	100	90.01
MPI	154550	100	100	100	100
MPV17	137960	100	100	100	94.91
MRPS16	609204	100	100	100	92.78
MRPS22	605810	100	100	100	94.60
MSMO1	607545	100	100	100	100
MTHFD1	172460	100	100	100	98.69
MTHFR	607093	100	100	99.74	95.92
MTHFS	604197	100	100	100	97.76
MTM1	300415	100	100	100	97.77
MTR	156570	100	100	99.44	97.37
MTRR	602568	100	100	100	99.45
MTTP	157147	100	100	100	99.79
MVK	251170	100	100	100	96.15
NAGA	104170	100	100	99.29	91.86
NAGLU	609701	100	94.32	91.15	85.27
NAGS	608300	100	100	99.31	89.81
NANS	605202	100	100	100	98.37
NAT8L	610647	86.18	75.10	72.57	64.80
NDUFA1	300078	100	100	100	100
NDUFA11	612638	100	100	99.93	83.25
NDUFA2	602137	100	100	100	100
NDUFAF1	606934	100	100	100	99.58
NDUFAF2	609653	100	100	97.27	79.30
NDUFAF3	612911	100	100	98.62	90.03
NDUFAF4	611776	100	100	100	92.65
NDUFAF5	612360	99.06	95.12	95.12	91.95
NDUFS1	157655	100	100	100	99.40
NDUFS2	602985	100	100	100	93.32
NDUFS3	603846	100	100	100	100
NDUFS4	602694	100	100	100	100
NDUFS6	603848	100	100	100	88.94
NDUFS7	601825	100	97.00	92.74	76.03
NDUFS8	602141	100	100	96.20	87.47
NDUFV1	161015	100	100	98.87	93.78
NDUFV2	600532	100	100	99.17	94.16

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
NEU1	608272	100	100	100	99.34
NFU1	608100	100	100	99.57	82.32
NHLRC1	608072	100	100	100	99.83
NPC1	607623	100	99.98	98.96	97.40
NPC2	601015	100	100	100	100
NPRL2	607072	100	100	100	98.42
NR5A2	604453	100	100	100	97.50
NSDHL	300275	100	100	100	99.92
NT5C	191720	99.86	99.86	95.38	63.44
NT5C3A	606224	100	100	97.48	89.44
NT5E	129190	100	100	100	99.53
NUBPL	613621	100	100	100	100
NUS1	610463	100	100	99.95	88.87
OAT	613349	100	96.42	93.55	89.43
OCRL	300535	98.15	98.15	98.15	97.22
OGDH	613022	100	100	100	99.89
OPA1	605290	100	100	98.77	92.07
OPA3	606580	100	100	100	100
OPLAH	614243	99.91	99.86	99.23	95.45
OTC	300461	100	100	100	96.95
OXCT1	601424	97.89	97.89	97.89	96.60
PAH	612349	100	100	100	100
PANK2	606157	100	100	100	99.69
PC	608786	100	100	99.95	96.64
PCBD1	126090	100	97.45	94.13	94.13
PCCA	232000	100	99.47	96.88	92.92
PCCB	232050	100	100	100	97.12
PCK1	614168	100	100	100	97.41
PCK2	614095	100	99.86	96.46	89.41
PDHA1	300502	100	100	98.88	92.65
PDHB	179060	100	100	100	99.37
PDHX	608769	100	100	99.58	84.45
PDK1	602524	100	100	100	98.91
PDK2	602525	100	100	98.15	92.02
PDK3	300906	100	100	100	100
PDK4	602527	100	100	100	99.90
PDP1	605993	100	100	100	100
PDP2	615499	100	100	100	100
PDSS1	607429	100	100	98.86	97.14
PDSS2	610564	100	100	98.78	93.22
PDXK	179020	100	100	100	99.48
PEPD	613230	100	100	98.93	91.34
PEX1	602136	100	99.82	97.75	93.52
PEX10	602859	100	100	100	94.29

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PEX11B	603867	100	100	100	89.09
PEX12	601758	100	100	100	100
PEX13	601789	100	100	100	96.20
PEX14	601791	100	100	98.17	83.71
PEX16	603360	100	100	95.63	67.73
PEX19	600279	100	100	95.14	86.96
PEX2	170993	100	100	100	100
PEX26	608666	100	100	100	95.81
PEX3	603164	100	100	100	99.12
PEX5	600414	100	100	99.73	93.66
PEX6	601498	100	99.12	95.44	87.85
PEX7	601757	100	100	99.06	87.85
PFKM	610681	100	100	100	99.98
PGAM2	612931	100	100	99.51	92.74
PGAP1	611655	100	99.36	95.40	86.16
PGAP2	615187	100	100	100	97.99
PGAP3	611801	100	100	100	100
PGK1	311800	100	100	100	95.92
PGM1	171900	100	100	100	97.45
PGM3	172100	100	100	100	100
PHGDH	606879	100	100	99.78	94.15
PHKA1	311870	100	100	99.91	96.54
PHKA2	300798	100	99.75	97.35	91.48
PHKB	172490	100	99.45	97.51	96.55
PHKG1	172470	100	100	98.42	94.34
PHKG2	172471	100	100	100	100
PHYH	602026	100	89.33	89.33	83.03
PIGA	311770	100	100	99.88	96.12
PIGC	601730	100	100	100	100
PIGG	616918	100	100	99.28	96.29
PIGL	605947	100	100	100	100
PIGM	610273	100	100	100	100
PIGN	606097	100	99.91	96.41	82.97
PIGO	614730	100	99.39	97.26	91.81
PIGQ	605754	100	100	99.02	93.29
PIGT	610272	100	100	98.63	89.87
PIGV	610274	100	100	100	100
PIGW	610275	100	100	100	100
PIGY	610662	100	100	100	100
PIK3R1	171833	100	100	100	98.75
PIK3R5	611317	100	100	99.05	89.70
PKLR	609712	100	100	100	98.43
PLA2G6	603604	100	100	99.56	94.14
PLCB1	607120	100	100	99.88	95.98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PLIN1	170290	100	98.03	89.83	65.15
PLOD1	153454	100	99.25	95.92	85.89
PLOD2	601865	96.78	95.59	91.29	86.31
PLOD3	603066	100	97.38	93.62	79.16
PLPBP	604436	100	100	91.78	87.92
PMM2	601785	100	100	100	98.16
PMPCA	613036	100	100	99.24	93.13
PNLIP	246600	100	100	100	98.26
PNP	164050	100	100	100	100
PNPLA2	609059	100	100	96.07	81.47
PNPLA6	603197	100	99.11	95.89	89.98
PNPO	603287	100	100	100	99.30
POFUT1	607491	100	100	100	88.49
POGLUT1	615618	100	100	100	100
POLG	174763	100	100	98.60	90.96
POLG2	604983	100	99.91	98.39	94.77
POLR3A	614258	100	100	100	99.12
POLR3B	614366	100	100	100	98.37
POMGNT1	606822	100	100	100	98.99
POMGNT2	614828	100	100	100	100
POMK	615247	100	100	100	100
POMT1	607423	100	100	99.08	95.04
POMT2	607439	100	100	99.89	98.71
POR	124015	96.93	96.93	96.93	96.64
PPM1K	611065	100	100	100	98.14
PPOX	600923	100	100	99.46	93.18
PPT1	600722	100	100	100	96.71
PRKAG2	602743	100	97.14	97.14	91.05
PRKCSH	177060	100	100	100	99.79
PRODH	606810	99.23	96.85	94.69	87.40
PRPS1	311850	100	100	100	99.75
PSAP	176801	100	100	100	95.51
PSAT1	610936	100	100	100	96.43
PSPH	172480	100	98.52	93.81	76.13
PTS	612719	100	100	100	100
PUS1	608109	100	100	99.93	91.65
PYCR1	179035	100	100	100	94.00
PYGL	613741	100	100	99.49	95.61
PYGM	608455	100	100	99.56	94.34
QDPR	612676	100	100	99.77	89.68
RARS2	611524	100	100	100	98.90
RBCK1	610924	100	100	100	91.33
RBP4	180250	100	100	100	100
RBSN	609511	100	100	100	97.49

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
RFT1	611908	100	99.84	98.65	91.37
RNASEH2A	606034	100	100	99.81	92.38
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
RNASET2	612944	100	100	96.99	89.77
RPIA	180430	100	100	100	100
RRM2B	604712	100	100	100	100
RXYLT1	605862	100	100	100	100
SAMHD1	606754	100	100	100	100
SAR1B	607690	100	96.36	87.96	87.96
SARDH	604455	100	100	99.27	93.45
SC5D	602286	100	100	100	100
SCARB2	602257	100	100	100	99.01
SCN3A	182391	100	100	99.88	99.25
SCO1	603644	100	100	100	100
SCO2	604272	100	100	100	100
SCP2	184755	100	100	100	100
SDHA	600857	100	97.10	96.38	94.50
SDHAF1	612848	100	100	100	86.16
SDHB	185470	100	100	100	99.60
SDHC	602413	100	100	100	100
SDHD	602690	100	100	100	100
SEC23B	610512	100	100	97.65	95.47
SEPSECS	613009	100	100	100	99.65
SERAC1	614725	100	100	100	100
SGSH	605270	93.52	93.52	93.52	93.25
SHMT1	182144	100	100	100	98.50
SHMT2	138450	100	100	100	95.92
SI	609845	100	99.60	97.21	92.96
SLC12A3	600968	100	99.56	98.20	90.28
SLC16A1	600682	100	100	100	100
SLC17A5	604322	93.58	93.58	93.58	92.55
SLC19A1	600424	100	100	99.95	98.32
SLC19A2	603941	100	100	100	94.48
SLC19A3	606152	99.94	99.94	99.94	99.94
SLC1A1	133550	100	100	100	98.81
SLC1A3	600111	100	100	100	91.40
SLC22A5	603377	100	100	100	98.98
SLC23A2	603791	100	100	100	98.47
SLC25A1	190315	100	100	100	100
SLC25A12	603667	100	100	100	99.62
SLC25A13	603859	100	100	100	98.07
SLC25A15	603861	100	100	100	100
SLC25A19	606521	100	100	99.40	96.48

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
SLC25A2	608157	100	100	100	100
SLC25A20	613698	100	100	100	100
SLC25A22	609302	100	100	100	98.43
SLC25A3	600370	100	100	100	99.96
SLC25A32	138480	100	100	100	100
SLC25A38	610819	100	100	100	100
SLC25A4	103220	100	100	99.59	94.76
SLC2A1	138140	100	97.73	97.73	97.73
SLC2A2	138160	100	100	100	99.33
SLC30A10	611146	100	100	100	100
SLC33A1	603690	100	100	100	99.18
SLC35A1	605634	100	100	96.30	95.28
SLC35A3	605632	100	100	100	100
SLC35C1	605881	100	100	100	99.65
SLC35D1	610804	100	100	99.00	91.53
SLC36A2	608331	100	99.64	96.09	84.96
SLC37A4	602671	99.87	99.87	99.42	96.62
SLC39A14	608736	97.92	97.92	97.03	91.16
SLC39A4	607059	100	100	100	100
SLC39A8	608732	100	100	100	98.70
SLC3A1	104614	100	100	100	98.87
SLC40A1	604653	100	100	99.68	97.86
SLC46A1	611672	99.87	99.87	99.87	99.77
SLC52A1	607883	100	100	98.14	92.42
SLC52A2	607882	100	100	100	100
SLC52A3	613350	100	100	99.46	96.97
SLC5A1	182380	100	100	100	96.95
SLC5A2	182381	100	100	100	99.87
SLC6A19	608893	100	98.37	91.99	82.21
SLC6A8	300036	99.70	91.39	77.18	53.90
SLC7A7	603593	100	100	100	99.01
SLC7A9	604144	100	100	100	98.91
SLCO1B1	604843	100	99.58	99.45	96.24
SLCO1B3	605495	100	100	99.83	94.34
SMPD1	607608	100	100	100	98.58
SOD1	147450	100	100	100	100
SPR	182125	100	100	99.05	88.20
SPTLC1	605712	100	100	100	100
SPTLC2	605713	100	100	98.91	94.83
SRD5A3	611715	100	100	98.58	94.73
SSR3	606213	100	100	100	100
SSR4	300090	100	100	100	99.57
ST3GAL3	606494	100	100	100	100
ST3GAL5	604402	100	100	100	99.37

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
STAR	600617	100	100	100	98.64
STS	300747	100	100	100	97.03
STT3A	601134	100	100	100	100
STT3B	608605	100	97.75	94.67	90.87
SUCLA2	603921	100	100	100	99.50
SUCLG1	611224	100	100	100	99.01
SUGCT	609187	100	100	100	97.61
SUMF1	607939	100	100	100	100
SUOX	606887	100	100	100	96.61
SURF1	185620	100	100	100	97.85
TACO1	612958	100	100	98.39	85.82
TFAZZIN	300394	-	-	-	-
TALDO1	602063	100	100	99.66	93.42
TANGO2	616830	100	100	99.95	96.86
TAT	613018	100	100	100	100
TBXAS1	274180	100	100	100	100
TCIRG1	604592	100	98.97	94.86	86.76
TCN1	189905	100	100	100	99.32
TCN2	613441	100	100	100	95.04
TDO2	191070	100	100	99.86	96.02
TECR	610057	100	99.67	87.71	65.66
TFR2	604720	100	100	97.45	84.53
TH	191290	100	98.04	94.21	86.67
THAP11	609119	100	100	100	100
TIMM8A	300356	100	100	100	93.05
TK2	188250	100	100	100	98.49
TMEM165	614726	100	100	100	96.75
TMEM199	616815	100	100	100	95.83
TMEM70	612418	100	100	97.50	90.77
TPI1	190450	100	97.80	94.69	90.48
TPMT	187680	100	100	100	100
TPP1	607998	100	98.87	96.77	93.28
TRAPPC11	614138	100	100	99.45	95.65
TREH	275360	100	99.01	94.94	92.91
TREX1	606609	100	100	100	100
TRIM37	605073	100	100	100	97.76
TRMU	610230	100	100	100	100
TRPM6	607009	100	99.77	98.46	96.42
TSFM	604723	100	100	100	100
TTC19	613814	100	100	100	97.45
TTPA	600415	99.89	98.72	94.97	79.66
TUFM	602389	100	100	100	99.74
TUSC3	601385	100	100	100	99.80
TWNK	606075	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
TYMP	131222	100	100	97.96	83.49
TYMS	188350	100	100	100	96.81
TYR	606933	100	100	100	99.76
TYRP1	115501	100	100	100	100
UGT1A1	191740	100	100	100	99.56
UMOD	191845	92.94	92.94	92.94	92.35
UMPS	613891	100	100	100	99.68
UPB1	606673	100	100	100	100
UQCRB	191330	100	100	100	100
UQCRCQ	612080	100	100	100	100
UROC1	613012	100	100	100	96.35
UROD	613521	100	100	100	99.77
UROS	606938	100	98.36	91.03	84.10
USF1	191523	100	100	100	100
VKORC1	608547	100	100	97.78	75.42
VPS13B	607817	99.28	98.29	97.00	91.79
XDH	607633	100	100	99.93	97.83
XYLT1	608124	98.78	95.46	92.27	85.10
XYLT2	608125	94.49	94.49	94.49	90.39
YARS2	610957	100	100	100	100
ZFYVE19	619635	100	99.08	96.02	81.48
ZNF143	603433	100	100	100	97.99

- 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 ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x