

Whole Exome Sequencing

Gene package Metabolic disorders, version 6, 21-2-2020



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. 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AASS	605113	63	100	97	90
ABAT	137150	100	100	100	96
ABCA1	600046	92	100	100	97
ABCB11	603201	67	100	100	96
ABCB4	171060	69	100	99	92
ABCB7	300135	59	100	98	87
ABCD1	300371	86	84	78	75
ABCD4	603214	108	100	100	100
ABCG2	603756	69	100	99	92
ABCG5	605459	80	100	100	99
ABCG8	605460	149	100	98	95
ABHD12	613599	61	100	100	94
ABHD5	604780	74	100	100	98
ACACA	200350	73	100	100	95
ACAD8	604773	159	100	100	100
ACAD9	611103	95	100	100	100
ACADM	607008	66	100	100	96

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ACADS	606885	121	100	100	100
ACADSB	600301	67	100	100	94
ACADVL	609575	114	100	100	98
ACAT1	607809	71	100	100	93
ACAT2	100678	92	100	100	98
ACBD5	616618	85	100	99	89
ACO2	100850	146	100	98	94
ACOX1	609751	103	100	100	99
ACSF3	614245	149	100	100	100
ACY1	104620	109	100	100	100
ADA	608958	85	100	100	96
ADCY5	600293	111	98	95	93
ADK	102750	53	100	93	80
ADSL	608222	89	100	100	96
AGA	613228	73	100	100	96
AGK	610345	57	100	100	93
AGL	610860	72	100	100	95
AGPAT2	603100	142	100	100	96
AGPS	603051	61	100	99	90
AGXT	604285	133	100	100	100
AGXT2	612471	71	100	99	92
AHCY	180960	151	96	96	96
AIFM1	300169	55	100	96	82
AK1	103000	124	100	100	100
AK2	103020	68	100	100	97
AKR1D1	604741	64	100	100	97
ALAD	125270	104	100	100	100
ALAS2	301300	60	100	98	90
ALDH18A1	138250	80	100	100	98
ALDH1A3	600463	88	100	100	97
ALDH3A2	609523	64	100	99	93
ALDH4A1	606811	118	100	100	99
ALDH5A1	610045	66	100	97	91
ALDH6A1	603178	98	100	100	98
ALDH7A1	107323	69	100	100	91
ALDOA	103850	144	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ALDOB	612724	102	100	100	100
ALG1	605907	61	91	79	73
ALG11	613666	73	100	100	99
ALG12	607144	158	100	100	100
ALG13	300776	50	100	97	83
ALG14	612866	102	100	100	97
ALG2	607905	81	100	100	99
ALG3	608750	95	100	100	100
ALG6	604566	73	100	100	96
ALG8	608103	62	100	100	94
ALG9	606941	64	100	100	94
ALOX12B	603741	113	100	100	99
ALPL	171760	130	100	100	100
AMACR	604489	96	100	100	98
AMN	605799	89	100	93	83
AMPD1	102770	67	100	99	95
AMPD2	102771	142	100	100	100
AMT	238310	130	100	100	100
APOA1	107680	130	100	100	100
APOB	107730	159	100	100	100
APOC2	608083	91	100	100	100
APOE	107741	102	100	100	95
APRT	102600	81	100	100	100
APTX	606350	76	100	99	92
ARG1	608313	75	100	100	98
ARSA	607574	136	100	100	100
ARSB	611542	73	100	100	98
ASAH1	613468	67	100	100	93
ASL	608310	117	100	100	99
ASNS	108370	73	100	100	92
ASPA	608034	55	100	99	91
ASS1	603470	115	100	98	90
ATIC	601731	62	100	100	96
ATP5F1E	606153	88	100	100	100
ATP6AP1	300197	76	100	99	95
ATP6V0A2	611716	81	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ATP7A	300011	49	100	98	84
ATP7B	606882	99	100	100	99
ATP8B1	602397	70	100	100	95
ATPAF2	608918	73	100	100	100
AUH	600529	94	100	100	98
B3GALNT1	603094	89	100	100	100
B3GALNT2	610194	59	100	100	97
B3GALT6	615291	64	79	75	72
B3GAT3	606374	125	100	100	100
B3GLCT	610308	67	100	99	85
B4GALNT1	601873	111	100	100	100
B4GALT1	137060	95	100	100	100
B4GALT7	604327	126	100	100	98
BAAT	602938	73	100	100	87
BCKDHA	608348	160	100	100	100
BCKDHB	248611	64	100	99	94
BCO1	605748	74	100	100	97
BCS1L	603647	170	100	100	100
BHMT	602888	58	100	95	86
BLVRA	109750	78	100	100	97
BMP2	112261	123	100	100	99
BOLA3	613183	69	100	100	91
BPGM	613896	71	100	100	99
BTD	609019	89	100	100	100
C1GALT1C1	300611	47	100	100	95
CA5A	114761	175	100	100	100
CAD	114010	121	100	100	100
CANT1	613165	128	100	100	100
CAT	115500	70	100	100	93
CBLIF	609342	No coverage data			
CBS	613381	127	100	100	100
CCDC115	613734	65	87	82	79
CD320	606475	95	100	100	100
CEL	114840	151	94	90	87
CERKL	608381	73	100	99	94
CERS3	615276	65	100	100	94

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CETP	118470	95	100	100	100
CHKB	612395	111	100	100	100
CHST14	608429	133	100	98	97
CHST3	603799	129	100	100	100
CHST6	605294	246	100	100	100
CHSY1	608183	79	98	95	92
CIC	612082	134	100	100	99
CLDN16	603959	73	100	100	97
CLDN19	610036	149	100	100	100
CLN3	607042	111	100	100	100
CLN5	608102	70	100	100	100
CLN6	606725	120	100	100	99
CLN8	607837	129	100	100	100
CLPB	616254	114	100	100	100
CLPS	120105	114	100	100	100
CLPX	615611	55	100	98	85
CNDP1	609064	72	100	100	96
CNNM2	607803	161	100	100	99
COG1	606973	96	100	100	98
COG2	606974	65	100	100	93
COG4	606976	81	100	100	99
COG5	606821	66	100	99	91
COG6	606977	65	100	99	90
COG7	606978	83	100	100	98
COG8	606979	125	100	100	100
COQ2	609825	66	100	100	94
COQ8A	606980	129	100	100	100
COX10	602125	139	100	100	99
COX15	603646	65	100	98	92
COX6B1	124089	102	100	100	100
CP	117700	63	100	100	96
CPOX	612732	82	100	100	98
CPS1	608307	61	100	100	96
CPT1A	600528	109	100	100	98
CPT2	600650	106	100	100	99
CRPPA	614631	81	100	99	93

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CRTC1	607536	146	100	100	99
CTH	607657	76	100	100	98
CTNS	606272	118	100	100	100
CTSA	613111	124	100	100	100
CTSC	602365	75	100	100	97
CTSD	116840	137	100	100	100
CTSK	601105	65	100	100	98
CUBN	602997	82	100	100	96
CYB5R3	613213	127	100	100	100
CYP27A1	606530	131	100	100	100
CYP27B1	609506	122	100	100	100
CYP7A1	118455	69	100	100	98
CYP7B1	603711	67	100	100	98
D2HGDH	609186	140	100	100	100
DARS2	610956	63	100	100	97
DBH	609312	136	100	100	100
DBT	248610	115	100	100	99
DCXR	608347	165	100	100	100
DDC	107930	84	100	99	92
DDHD1	614603	107	100	98	92
DDOST	602202	92	100	100	100
DGAT1	604900	116	96	91	85
DGKE	601440	106	100	100	98
DGUOK	601465	73	100	100	92
DHCR24	606418	119	100	100	100
DHCR7	602858	112	100	100	100
DHDDS	608172	69	100	100	99
DHFR	126060	76	100	100	98
DHODH	126064	87	100	100	96
DHTKD1	614984	85	100	100	98
DLAT	608770	74	100	98	90
DLD	238331	77	100	100	98
DLST	126063	81	100	100	100
DMGDH	605849	75	100	100	99
DNAJC12	606060	69	100	95	88
DNAJC19	608977	75	100	100	96

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
DNAJC5	611203	217	100	100	100
DNM1L	603850	70	100	99	94
DNMT1	126375	93	100	100	97
DOLK	610746	136	100	100	100
DPAGT1	191350	76	100	100	100
DPEP1	179780	124	100	100	100
DPM1	603503	79	94	89	85
DPM2	603564	76	100	100	100
DPM3	605951	227	100	100	100
DPYD	612779	66	100	99	95
DPYS	613326	75	100	100	98
EBP	300205	93	100	100	100
ECHS1	602292	79	100	100	100
EGF	131530	74	100	100	97
ELOVL4	605512	71	100	100	98
ENO3	131370	148	100	100	100
EOGT	614789	60	100	100	94
EPG5	615068	69	100	100	97
EPHX1	132810	128	100	100	99
EPHX2	132811	88	100	100	95
EPM2A	607566	86	89	87	85
ETFA	608053	60	100	100	96
ETFB	130410	106	100	100	100
ETFDH	231675	85	100	100	99
ETHE1	608451	113	100	100	98
EXT1	608177	70	100	100	96
EXT2	608210	98	100	100	97
FA2H	611026	83	100	99	91
FAH	613871	99	100	100	100
FASTKD2	612322	74	100	100	96
FBP1	611570	119	100	100	100
FECH	612386	64	100	100	96
FGFR2	176943	74	100	100	95
FH	136850	82	99	94	87
FKRP	606596	143	100	100	100
FKTN	607440	85	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
FLAD1	610595	136	100	100	100
FMO3	136132	69	100	100	95
FOLR1	136430	111	100	100	100
FOXRED1	613622	91	100	100	100
FTCD	606806	96	99	96	93
FUCA1	612280	83	100	100	98
FUT2	182100	182	100	100	100
FXYD2	601814	101	100	100	100
G6PC	613742	113	100	100	100
G6PC3	611045	103	100	100	100
G6PD	305900	96	100	100	100
GAA	606800	156	100	100	100
GAD1	605363	86	100	100	99
GALC	606890	54	100	99	89
GALE	606953	116	100	100	100
GALK1	604313	133	100	100	99
GALNS	612222	81	100	99	95
GALNT3	601756	69	100	100	97
GALT	606999	151	100	100	100
GAMT	601240	85	100	99	95
GANAB	104160	85	100	99	96
GART	138440	55	100	96	89
GATM	602360	65	100	100	98
GBA	606463	172	100	100	100
GBA2	609471	137	100	100	100
GBE1	607839	67	100	99	92
GCDH	608801	112	100	100	100
GCH1	600225	52	100	100	93
GCK	138079	127	100	100	100
GCLC	606857	88	100	100	97
GCSH	238330	93	100	89	62
GFM1	606639	72	100	100	96
GFPT1	138292	59	100	100	92
GGT1	612346	54	77	63	57
GK	300474	46	99	88	66
GLA	300644	101	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GLB1	611458	105	100	100	98
GLDC	238300	67	100	98	91
GLRA1	138491	85	100	100	96
GLRX5	609588	68	100	91	81
GLUD1	138130	99	100	97	91
GLUL	138290	92	100	100	100
GLYCTK	610516	136	100	100	100
GM2A	613109	94	100	100	100
GMPPA	615495	114	100	100	100
GMPPB	615320	189	100	100	100
GNE	603824	93	100	100	99
GNMT	606628	105	100	100	100
GNPAT	602744	78	100	100	96
GNPTAB	607840	63	100	99	95
GNPTG	607838	153	100	99	94
GNS	607664	68	100	100	98
GPD1	138420	80	100	100	100
GPHN	603930	68	100	100	98
GPI	172400	130	100	100	100
GPX1	138320	95	100	100	100
GRHPR	604296	96	100	100	100
GSS	601002	89	100	100	99
GSTZ1	603758	95	100	100	100
GUSB	611499	106	100	100	97
GYG1	603942	60	100	100	96
GYS1	138570	116	100	100	100
GYS2	138571	67	100	100	98
HADH	601609	77	100	100	100
HADHA	600890	99	100	100	97
HADHB	143450	66	100	100	97
HAL	609457	89	100	100	97
HAMP	606464	161	100	100	100
HCFC1	300019	79	100	97	92
HEXA	606869	86	100	100	98
HEXB	606873	119	100	100	97
HFE	613609	97	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
HGD	607474	63	100	100	96
HGSNAT	610453	70	94	94	92
HIBCH	610690	51	100	94	75
HK1	142600	102	100	100	99
HLCS	609018	98	100	100	99
HMBS	609806	88	100	100	100
HMGCL	613898	94	100	100	100
HMGCS2	600234	78	100	100	99
HMOX1	141250	112	100	100	99
HOGA1	613597	122	100	100	100
HPD	609695	107	100	100	99
HPRT1	308000	43	100	99	81
HPS1	604982	103	100	100	97
HS6ST1	604846	119	100	100	97
HSD11B1	600713	68	100	100	99
HSD11B2	614232	147	92	86	83
HSD17B10	300256	76	100	100	100
HSD17B4	601860	62	100	98	93
HSD3B2	613890	143	100	100	100
HSD3B7	607764	115	100	100	100
HYAL1	607071	103	100	100	100
IBA57	615316	124	100	100	97
IDH2	147650	105	100	100	100
IDH3B	604526	117	100	100	100
IDS	300823	74	100	99	95
IDUA	252800	138	100	97	91
IMPAD1	614010	146	100	100	97
IMPDH1	146690	99	100	96	91
INPPL1	600829	111	100	100	99
ISCU	611911	75	100	100	100
ITPA	147520	122	100	100	100
IVD	607036	107	100	100	98
KHK	614058	119	100	100	100
KYNU	605197	73	100	100	97
L2HGDH	609584	74	100	100	97
LAMP2	309060	44	100	95	77

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
LARGE1	603590	97	100	100	99
LBR	600024	68	100	100	96
LCAT	606967	130	100	100	97
LCT	603202	114	100	100	100
LDHA	150000	62	100	99	88
LDLR	606945	209	100	100	100
LFNG	602576	129	86	84	83
LIPA	613497	68	100	100	96
LIPC	151670	82	100	100	98
LIPI	609252	60	100	99	92
LMBRD1	612625	65	100	98	85
LPIN1	605518	78	100	100	97
LPIN2	605519	77	100	100	98
LPL	609708	110	100	100	99
LRP2	600073	69	100	100	96
LRPPRC	607544	60	100	99	91
LTC4S	246530	80	100	85	74
LYST	606897	72	100	99	95
MAN1B1	604346	122	100	100	100
MAN2B1	609458	115	100	100	100
MANBA	609489	85	100	100	95
MAOA	309850	49	100	99	92
MAT1A	610550	107	100	100	100
MAT2A	601468	83	100	100	96
MCCC1	609010	74	100	100	94
MCCC2	609014	64	100	100	96
MCEE	608419	77	100	100	95
MCOLN1	605248	129	100	100	100
MFSD8	611124	68	100	100	95
MGAT2	602616	104	100	100	100
MLYCD	606761	65	100	96	92
MMAA	607481	78	100	100	98
MMAB	607568	82	100	100	98
MMACHC	609831	133	100	100	100
MMADHC	611935	63	100	100	90
MMUT	609058	75	100	100	95

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
MOCOS	613274	87	100	100	95
MOCS1	603707	105	100	100	99
MOCS2	603708	67	100	100	96
MOCS3	609277	176	100	100	100
MOGS	601336	130	100	100	100
MPDU1	604041	87	100	100	99
MPI	154550	154	100	100	99
MPV17	137960	108	100	100	100
MRPS16	609204	120	100	100	100
MRPS22	605810	75	100	100	96
MSMO1	607545	95	100	100	98
MTHFD1	172460	76	100	100	96
MTHFR	607093	99	100	100	99
MTHFS	604197	107	100	100	100
MTM1	300415	46	100	95	75
MTR	156570	80	100	100	97
MTRR	602568	87	100	100	98
MTPP	157147	67	100	100	97
MVK	251170	100	100	100	100
NAGA	104170	118	100	100	100
NAGLU	609701	112	100	97	93
NAGS	608300	110	100	100	100
NANS	605202	65	100	99	94
NAT8L	610647	73	94	90	84
NDUFA1	300078	110	100	100	100
NDUFA11	612638	117	100	100	100
NDUFA2	602137	104	100	100	100
NDUFAF1	606934	63	100	100	95
NDUFAF2	609653	83	100	92	78
NDUFAF3	612911	130	100	100	100
NDUFAF4	611776	95	100	100	100
NDUFAF5	612360	70	100	100	96
NDUFS1	157655	69	100	100	94
NDUFS2	602985	79	100	100	99
NDUFS3	603846	112	100	100	100
NDUFS4	602694	83	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NDUFS6	603848	70	100	100	100
NDUFS7	601825	130	100	100	98
NDUFS8	602141	140	100	100	100
NDUFV1	161015	129	100	100	100
NDUFV2	600532	48	100	95	80
NEU1	608272	155	100	100	100
NFU1	608100	66	100	100	95
NHLRC1	608072	144	100	100	100
NPC1	607623	87	100	100	99
NPC2	601015	95	100	100	100
NPRL2	607072	119	100	100	100
NR5A2	604453	81	100	100	98
NSDHL	300275	68	100	100	93
NT5C	191720	93	100	81	72
NT5C3A	606224	61	100	99	89
NT5E	129190	90	100	100	98
NUBPL	613621	60	100	100	97
NUS1	610463	54	100	98	78
OAT	613349	64	100	96	85
OCRL	300535	43	100	95	80
OGDH	613022	131	100	100	99
OPA1	605290	59	100	98	89
OPA3	606580	130	100	100	100
OPLAH	614243	132	100	100	99
OTC	300461	58	100	98	85
OXCT1	601424	74	100	100	96
PAH	612349	68	100	99	95
PANK2	606157	81	100	100	99
PC	608786	141	100	100	100
PCBD1	126090	87	100	100	99
PCCA	232000	68	100	100	95
PCCB	232050	72	100	98	95
PCK1	614168	114	100	100	100
PCK2	614095	121	100	100	100
PDHA1	300502	49	100	94	84
PDHB	179060	65	100	100	95

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PDHX	608769	79	100	100	97
PDK1	602524	74	100	100	96
PDK2	602525	117	100	100	100
PDK3	300906	52	100	98	82
PDK4	602527	57	100	100	93
PDP1	605993	87	100	100	100
PDP2	615499	126	100	100	100
PDSS1	607429	60	100	93	85
PDSS2	610564	75	100	97	87
PDXK	179020	98	100	100	100
PEPD	613230	100	100	100	98
PEX1	602136	62	100	99	94
PEX10	602859	100	100	100	97
PEX11B	603867	137	100	100	100
PEX12	601758	67	100	100	94
PEX13	601789	69	100	100	99
PEX14	601791	142	100	100	100
PEX16	603360	134	100	96	94
PEX19	600279	67	100	100	98
PEX2	170993	63	100	100	100
PEX26	608666	118	100	100	100
PEX3	603164	57	100	100	95
PEX5	600414	111	100	100	100
PEX6	601498	107	100	99	95
PEX7	601757	57	100	100	95
PFKM	610681	92	100	100	99
PGAM2	612931	156	100	100	100
PGAP1	611655	65	100	99	90
PGAP2	615187	147	100	100	100
PGAP3	611801	115	100	100	100
PGK1	311800	47	100	100	89
PGM1	171900	72	100	100	99
PGM3	172100	88	100	100	98
PHGDH	606879	138	100	100	100
PHKA1	311870	47	100	97	82
PHKA2	300798	56	100	99	90

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PHKB	172490	76	100	100	98
PHKG1	172470	123	100	100	97
PHKG2	172471	123	100	100	100
PHYH	602026	121	100	100	94
PIGA	311770	67	100	100	97
PIGC	601730	149	100	100	100
PIGG	616918	100	100	100	98
PIGL	605947	98	100	100	100
PIGM	610273	85	100	100	100
PIGN	606097	63	100	99	89
PIGO	614730	126	100	100	100
PIGQ	605754	157	100	100	100
PIGT	610272	138	100	100	100
PIGV	610274	96	100	100	100
PIGW	610275	85	100	100	100
PIGY	610662	44	100	100	94
PIK3R1	171833	80	100	100	97
PIK3R5	611317	107	100	100	100
PKLR	609712	156	100	100	100
PLA2G6	603604	118	100	100	99
PLCB1	607120	60	100	99	94
PLIN1	170290	96	100	100	99
PLOD1	153454	100	100	100	99
PLOD2	601865	58	100	99	90
PLOD3	603066	100	100	100	99
PLPBP	604436	53	100	97	84
PMM2	601785	72	100	100	97
PMPCA	613036	133	100	100	100
PNLIP	246600	60	100	100	95
PNP	164050	70	100	100	97
PNPLA2	609059	114	100	100	100
PNPLA6	603197	123	100	100	99
PNPO	603287	69	100	100	94
POFUT1	607491	114	100	100	100
POGLUT1	615618	54	100	98	91
POLG	174763	114	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
POLG2	604983	140	100	100	95
POLR3A	614258	84	100	100	98
POLR3B	614366	76	100	99	93
POMGNT1	606822	93	100	100	99
POMGNT2	614828	154	100	100	100
POMK	615247	95	100	100	100
POMT1	607423	103	100	100	100
POMT2	607439	87	100	100	99
POR	124015	170	100	100	100
PPM1K	611065	77	100	100	98
PPOX	600923	106	100	100	100
PPT1	600722	84	100	100	100
PRKAG2	602743	123	100	100	97
PRKCSH	177060	142	100	100	98
PRODH	606810	98	100	95	86
PRPS1	311850	50	100	99	90
PSAP	176801	121	100	100	97
PSAT1	610936	64	100	100	97
PSPH	172480	51	100	96	84
PTS	612719	86	100	100	95
PUS1	608109	88	100	99	95
PYCR1	179035	93	100	100	100
PYGL	613741	84	100	100	96
PYGM	608455	112	100	100	100
QDPR	612676	94	100	100	97
RARS2	611524	66	100	99	93
RBCK1	610924	135	100	100	99
RBP4	180250	128	100	100	100
RBSN	609511	110	100	100	97
RFT1	611908	67	100	99	93
RNASEH2A	606034	109	100	100	100
RNASEH2B	610326	62	100	98	88
RNASEH2C	610330	323	100	100	100
RNASET2	612944	106	100	100	99
RPIA	180430	77	100	100	98
RRM2B	604712	90	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RXYLT1	605862	90	100	100	94
SAMHD1	606754	64	100	99	88
SAR1B	607690	79	100	100	97
SARDH	604455	104	100	100	99
SC5D	602286	88	100	100	100
SCARB2	602257	89	100	100	98
SCN3A	182391	84	100	100	97
SCO1	603644	99	100	100	99
SCO2	604272	125	100	100	100
SCP2	184755	54	100	100	92
SDHA	600857	134	100	97	91
SDHAF1	612848	82	100	100	100
SDHB	185470	149	100	100	100
SDHC	602413	170	100	100	100
SDHD	602690	164	100	100	100
SEC23B	610512	74	100	100	97
SEPSECS	613009	78	100	100	96
SERAC1	614725	65	100	99	86
SGSH	605270	103	100	94	90
SHMT1	182144	95	100	100	100
SHMT2	138450	135	100	100	99
SI	609845	60	100	99	92
SLC12A3	600968	113	100	100	100
SLC16A1	600682	83	100	100	99
SLC17A5	604322	86	100	100	97
SLC19A1	600424	167	100	99	97
SLC19A2	603941	68	100	100	99
SLC19A3	606152	73	100	100	98
SLC1A1	133550	93	100	100	98
SLC1A3	600111	99	100	100	100
SLC22A5	603377	118	100	100	100
SLC23A2	603791	74	100	99	95
SLC25A1	190315	102	100	100	100
SLC25A12	603667	88	100	100	97
SLC25A13	603859	98	100	99	93
SLC25A15	603861	132	100	100	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC25A19	606521	105	100	100	100
SLC25A20	613698	66	100	100	97
SLC25A22	609302	122	100	100	100
SLC25A3	600370	105	100	100	100
SLC25A32	610815	58	100	97	83
SLC25A38	610819	109	100	100	100
SLC25A4	103220	112	100	100	100
SLC2A1	138140	127	100	100	100
SLC2A2	138160	63	100	100	96
SLC30A10	611146	148	100	100	100
SLC33A1	603690	68	100	98	86
SLC35A1	605634	66	100	100	97
SLC35A3	605632	73	100	99	92
SLC35C1	605881	146	100	100	100
SLC35D1	610804	57	100	99	85
SLC36A2	608331	102	100	100	100
SLC37A4	602671	96	100	100	95
SLC39A14	608736	92	100	100	97
SLC39A4	607059	125	100	100	100
SLC39A8	608732	83	100	99	95
SLC3A1	104614	93	100	100	97
SLC40A1	604653	63	100	100	97
SLC46A1	611672	121	100	100	98
SLC52A1	607883	203	100	100	100
SLC52A2	607882	173	100	100	100
SLC52A3	613350	109	100	100	100
SLC5A1	182380	104	100	100	96
SLC5A2	182381	113	100	100	99
SLC6A19	608893	115	100	100	100
SLC6A8	300036	103	100	98	95
SLC7A7	603593	79	100	100	97
SLC7A9	604144	100	100	100	99
SLCO1B1	604843	61	100	99	90
SLCO1B3	605495	66	100	99	92
SMPD1	607608	131	100	100	99
SOD1	147450	74	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SPR	182125	102	100	100	100
SPTLC1	605712	62	100	99	94
SPTLC2	605713	81	100	100	96
SRD5A3	611715	111	100	100	96
SSR3	606213	63	94	77	57
SSR4	300090	76	100	100	99
ST3GAL3	606494	79	100	100	96
ST3GAL5	604402	55	99	92	84
STAR	600617	118	100	100	100
STS	300747	90	97	97	95
STT3A	601134	63	100	100	98
STT3B	608605	90	100	100	97
SUCLA2	603921	56	100	97	86
SUCLG1	611224	88	100	100	100
SUGCT	609187	73	100	100	93
SUMF1	607939	88	100	100	99
SUOX	606887	136	100	100	100
SURF1	185620	88	94	89	87
TACO1	612958	82	100	100	100
TALDO1	602063	136	100	100	100
TANGO2	616830	96	100	100	99
TAT	613018	65	100	100	98
TAZ	300394	97	100	98	90
TBXAS1	274180	74	100	99	93
TCIRG1	604592	117	100	100	97
TCN1	189905	66	100	99	95
TCN2	613441	118	100	100	100
TDO2	191070	49	100	99	88
TECR	610057	140	100	100	100
TFR2	604720	101	100	100	97
TH	191290	93	100	99	94
TIMM8A	300356	138	100	100	100
TK2	188250	66	100	100	95
TMEM165	614726	108	100	100	100
TMEM199	616815	85	100	100	100
TMEM70	612418	93	100	100	95

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TPI1	190450	123	100	98	96
TPMT	187680	75	100	100	89
TPP1	607998	97	100	100	100
TRAPPC11	614138	60	100	97	90
TREH	275360	111	100	100	100
TREX1	606609	234	100	100	100
TRIM37	605073	61	100	100	94
TRMU	610230	97	100	100	98
TRPM6	607009	75	100	100	96
TSFM	604723	77	100	100	100
TTC19	613814	53	100	92	77
TTPA	600415	61	100	98	91
TUFM	602389	158	100	100	100
TUSC3	601385	72	100	100	95
TWNK	606075	145	100	100	100
TYMP	131222	102	100	100	100
TYMS	188350	76	100	100	97
TYR	606933	83	100	100	98
TYRP1	115501	73	100	100	98
UGT1A1	191740	130	100	100	100
UMOD	191845	95	100	100	97
UMPS	613891	74	100	100	98
UPB1	606673	96	100	100	100
UQCRB	191330	75	100	100	100
UQCRQ	612080	123	100	100	100
UROC1	613012	112	100	100	99
UROD	613521	87	100	100	97
UROS	606938	60	100	100	97
USF1	191523	113	100	100	100
VKORC1	608547	113	100	100	99
VPS13B	607817	75	100	99	96
XDH	607633	81	100	100	96
XYLT1	608124	103	100	97	93
XYLT2	608125	131	100	100	100
YARS2	610957	127	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
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- 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 (± 10 bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (± 10 bp flanking introns) that is covered at least 10x, 20x or 30x