

Whole Exome Sequencing

Gene package Metabolic disorders, version 7, 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. 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	87	100	98	93
ABAT	137150	142	100	100	100
ABCA1	600046	128	100	100	99
ABCB11	603201	114	100	99	97
ABCB4	171060	98	98	96	91
ABCB7	300135	64	100	94	83
ABCD1	300371	178	100	100	100
ABCD4	603214	169	100	100	100
ABCG2	603756	79	100	100	98
ABCG5	605459	115	100	99	91
ABCG8	605460	188	100	100	100
ABHD12	613599	133	100	99	93
ABHD5	604780	116	100	96	92
ACACA	200350	97	99	99	97
ACAD8	604773	186	100	100	100
ACAD9	611103	186	100	100	100
ACADM	607008	51	93	80	68
ACADS	606885	349	100	100	100
ACADSB	600301	74	100	99	94
ACADVL	609575	238	100	100	100
ACAT1	607809	65	96	91	80
ACAT2	100678	156	100	100	100
ACBD5	616618	104	100	100	97
ACO2	100850	270	100	100	100
ACOX1	609751	126	100	100	98
ACSF3	614245	244	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ACY1	104620	215	100	100	100
ADA	608958	161	100	100	100
ADCY5	600293	259	100	99	98
ADK	102750	58	99	89	81
ADSL	608222	178	100	100	100
AGA	613228	83	100	98	90
AGK	610345	97	100	97	97
AGL	610860	68	98	91	83
AGPAT2	603100	227	100	100	100
AGPS	603051	64	96	94	85
AGXT	604285	248	100	100	100
AGXT2	612471	118	100	100	97
AHCY	180960	268	100	100	100
AIFM1	300169	89	100	99	96
AK1	103000	147	100	97	94
AK2	103020	99	100	100	98
AKR1D1	604741	87	100	100	97
ALAD	125270	217	100	100	100
ALAS2	301300	102	100	100	100
ALDH18A1	138250	169	100	100	99
ALDH1A3	600463	129	100	100	99
ALDH3A2	609523	90	100	100	97
ALDH4A1	606811	315	100	100	100
ALDH5A1	610045	121	100	100	98
ALDH6A1	603178	116	100	100	99
ALDH7A1	107323	105	100	100	95
ALDOA	103850	278	100	100	100
ALDOB	612724	127	100	100	100
ALG1	605907	126	100	100	99
ALG11	613666	109	100	100	95
ALG12	607144	224	100	100	100
ALG13	300776	67	98	92	81
ALG14	612866	108	100	100	100
ALG2	607905	173	100	100	100
ALG3	608750	213	100	100	100
ALG6	604566	49	90	81	67
ALG8	608103	62	93	86	79
ALG9	606941	78	97	95	92
ALOX12B	603741	262	100	100	100
ALPL	171760	323	100	100	100
AMACR	604489	129	100	100	99
AMN	605799	200	100	100	100
AMPD1	102770	90	100	99	95
AMPD2	102771	219	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AMT	238310	225	100	100	100
APOA1	107680	213	100	100	100
APOB	107730	309	100	100	99
APOC2	608083	138	100	100	100
APOE	107741	151	100	100	100
APRT	102600	203	100	100	100
APTX	606350	166	100	98	90
ARG1	608313	89	100	99	95
ARSA	607574	380	100	100	100
ARSB	611542	130	100	100	98
ASAH1	613468	73	98	92	81
ASL	608310	294	100	100	100
ASNS	108370	82	100	98	91
ASPA	608034	103	100	98	94
ASS1	603470	215	100	98	96
ATIC	601731	84	100	99	94
ATP5F1E	606153	95	100	100	100
ATP6AP1	300197	134	100	100	99
ATP6V0A2	611716	120	100	97	91
ATP7A	300011	59	99	94	81
ATP7B	606882	195	100	100	100
ATP8B1	602397	84	99	92	86
ATPAF2	608918	175	100	100	97
AUH	600529	81	100	98	91
B3GALNT1	603094	109	100	100	99
B3GALNT2	610194	101	100	100	99
B3GALT6	615291	283	83	80	78
B3GAT3	606374	243	100	100	100
B3GLCT	610308	64	97	84	73
B4GALNT1	601873	269	100	100	100
B4GALT1	137060	136	100	100	100
B4GALT7	604327	254	100	100	100
BAAT	602938	243	100	100	99
BCKDHA	608348	221	100	100	100
BCKDHB	248611	137	100	100	99
BCKDK	614901	220	100	100	100
BCO1	605748	129	100	100	100
BCS1L	603647	263	100	100	100
BHMT	602888	99	100	97	90
BLVRA	109750	138	100	100	100
BMP2	112261	211	100	99	98
BOLA3	613183	63	100	98	94
BPGM	613896	117	100	100	99
BTD	609019	195	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
C1GALT1C1	300611	166	100	100	100
CA5A	114761	147	100	100	100
CAD	114010	218	100	100	100
CANT1	613165	318	100	100	100
CAT	115500	120	100	100	100
CBLIF	609342	No coverage data			
CBS	613381	430	100	100	100
CCDC115	613734	184	100	100	100
CD320	606475	320	100	100	100
CEL	114840	254	97	95	91
CERKL	608381	82	100	94	81
CERS3	615276	66	100	100	92
CETP	118470	174	100	100	100
CHKB	612395	373	100	100	100
CHST14	608429	278	100	100	98
CHST3	603799	345	100	100	100
CHST6	605294	533	100	100	100
CHSY1	608183	141	97	94	94
CIC	612082	232	100	100	100
CLDN16	603959	120	100	100	100
CLDN19	610036	369	100	100	100
CLN3	607042	211	100	100	100
CLN5	608102	113	100	93	89
CLN6	606725	264	100	100	100
CLN8	607837	195	100	100	100
CLPB	616254	209	100	100	100
CLPS	120105	247	100	100	100
CLPX	615611	81	100	99	94
CNDP1	609064	109	100	100	98
CNNM2	607803	209	100	100	97
COG1	606973	164	100	100	99
COG2	606974	81	96	92	89
COG4	606976	150	100	100	100
COG5	606821	73	95	92	85
COG6	606977	66	94	83	74
COG7	606978	172	100	100	100
COG8	606979	164	100	100	100
COQ2	609825	79	98	92	82
COQ8A	606980	250	100	100	100
COX10	602125	177	100	99	94
COX15	603646	137	100	100	100
COX6B1	124089	158	100	100	100
CP	117700	92	99	98	94
CPOX	612732	103	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CPS1	608307	101	100	100	99
CPT1A	600528	163	100	100	99
CPT2	600650	187	100	100	100
CRPPA	614631	76	97	84	77
CRTC1	607536	206	100	100	100
CTH	607657	91	100	98	92
CTNS	606272	147	100	97	90
CTSA	613111	181	100	100	100
CTSC	602365	182	100	100	100
CTSD	116840	278	100	100	100
CTSK	601105	108	100	100	100
CUBN	602997	107	100	99	96
CYB5R3	613213	188	100	100	100
CYP27A1	606530	295	100	100	100
CYP27B1	609506	256	100	100	100
CYP7A1	118455	117	100	100	99
CYP7B1	603711	111	100	99	94
D2HGDH	609186	269	100	100	100
DARS2	610956	79	100	96	94
DBH	609312	256	100	100	100
DBT	248610	70	100	93	83
DCXR	608347	314	100	100	100
DDC	107930	112	100	95	92
DDHD1	614603	143	100	100	97
DDOST	602202	211	100	100	100
DGAT1	604900	341	99	96	94
DGKE	601440	149	89	85	81
DGUOK	601465	115	100	100	100
DHCR24	606418	207	100	100	100
DHCR7	602858	243	100	100	100
DHDDS	608172	146	100	100	100
DHFR	126060	57	93	71	43
DHODH	126064	225	100	100	99
DHTKD1	614984	128	100	100	99
DLAT	608770	131	100	99	92
DLD	238331	65	99	90	76
DLST	126063	109	100	99	96
DMGDH	605849	87	100	96	92
DNAJC12	606060	97	100	98	92
DNAJC19	608977	70	100	98	85
DNAJC5	611203	258	100	100	100
DNM1L	603850	61	99	92	82
DNMT1	126375	192	100	100	98
DOLK	610746	430	100	100	100

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DPAGT1	191350	151	100	100	100
DPEP1	179780	275	100	100	100
DPM1	603503	79	98	89	73
DPM2	603564	164	100	100	100
DPM3	605951	175	100	100	100
DPYD	612779	73	94	92	86
DPYS	613326	160	100	100	98
EBP	300205	110	100	100	100
ECHS1	602292	170	100	100	100
EGF	131530	110	99	95	91
ELOVL4	605512	98	98	89	81
ENO3	131370	236	100	100	100
EOGT	614789	69	100	95	90
EPG5	615068	106	100	99	98
EPHX1	132810	136	100	100	98
EPHX2	132811	130	100	100	99
EPM2A	607566	164	90	86	84
ETFA	608053	77	100	92	83
ETFB	130410	179	100	100	100
ETFDH	231675	73	100	92	81
ETHE1	608451	197	100	100	100
EXT1	608177	298	100	100	100
EXT2	608210	142	100	100	100
FA2H	611026	182	100	100	100
FAH	613871	186	100	100	100
FASTKD2	612322	72	100	98	91
FBP1	611570	229	100	100	100
FECH	612386	139	100	100	99
FGFR2	176943	145	100	100	99
FH	136850	83	100	100	98
FKRP	606596	339	100	100	100
FKTN	607440	104	90	81	68
FLAD1	610595	269	100	100	100
FMO3	136132	157	100	100	99
FOLR1	136430	146	100	100	100
FOXRED1	613622	260	100	100	100
FTCD	606806	270	100	99	98
FUCA1	612280	172	100	100	100
FUT2	182100	243	100	100	100
FXD2	601814	241	100	100	100
G6PC	613742	178	100	100	100
G6PC3	611045	161	100	99	95
G6PD	305900	211	100	100	100
GAA	606800	259	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GAD1	605363	139	100	100	99
GALC	606890	85	98	94	88
GALE	606953	200	100	100	100
GALK1	604313	319	100	100	100
GALNS	612222	220	100	100	100
GALNT3	601756	74	99	89	82
GALT	606999	231	100	100	100
GAMT	601240	284	100	100	100
GANAB	104160	156	100	100	100
GART	138440	104	100	100	99
GATM	602360	105	100	100	99
GBA	606463	207	100	100	100
GBA2	609471	212	100	100	100
GBE1	607839	87	100	100	98
GCDH	608801	239	100	100	100
GCH1	600225	207	94	94	94
GCK	138079	256	100	100	100
GCLC	606857	103	100	96	91
GCSH	238330	43	100	85	69
GFM1	606639	82	100	99	94
GFPT1	138292	90	100	100	97
GGT1	612346	97	100	91	80
GK	300474	27	83	59	35
GLA	300644	87	100	100	98
GLB1	611458	149	100	100	100
GLDC	238300	118	100	100	98
GLRA1	138491	154	100	100	100
GLRX5	609588	125	100	100	100
GLUD1	138130	123	100	98	93
GLUL	138290	140	100	100	100
GLYCTK	610516	298	100	100	100
GM2A	613109	183	100	100	100
GMPPA	615495	180	100	100	100
GMPPB	615320	281	100	100	100
GNE	603824	123	100	100	97
GNMT	606628	215	100	100	100
GNPAT	602744	80	97	94	86
GNPTAB	607840	87	100	99	95
GNPTG	607838	302	100	100	100
GNS	607664	105	100	100	99
GPD1	138420	219	100	100	100
GPHN	603930	87	100	96	94
GPI	172400	217	100	100	100
GPX1	138320	310	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GRHPR	604296	185	100	100	100
GSS	601002	188	100	100	100
GSTZ1	603758	176	100	100	100
GUSB	611499	222	100	100	100
GYG1	603942	107	100	100	98
GYS1	138570	231	100	100	100
GYS2	138571	96	99	94	89
HADH	601609	145	100	100	98
HADHA	600890	125	100	99	95
HADHB	143450	74	90	84	76
HAL	609457	182	100	99	96
HAMP	606464	154	100	100	100
HCFC1	300019	207	100	100	100
HEXA	606869	162	100	100	100
HEXB	606873	73	97	91	84
HFE	613609	175	100	100	100
HGD	607474	123	100	100	98
HGSNAT	610453	93	99	94	92
HIBCH	610690	68	95	91	80
HK1	142600	158	100	100	99
HLCS	609018	163	100	100	99
HMBS	609806	202	100	100	100
HMGCL	613898	168	100	100	100
HMGCS2	600234	176	100	100	99
HMOX1	141250	187	100	100	100
HOGA1	613597	229	100	100	100
HPD	609695	196	100	100	100
HPRT1	308000	40	95	80	55
HPS1	604982	250	100	100	100
HS6ST1	604846	330	100	100	100
HSD11B1	600713	111	100	100	97
HSD11B2	614232	232	95	92	86
HSD17B10	300256	123	100	100	100
HSD17B4	601860	78	93	91	84
HSD3B2	613890	170	100	100	100
HSD3B7	607764	275	100	100	100
HYAL1	607071	247	100	100	100
IBA57	615316	227	100	100	100
IDH2	147650	236	100	100	100
IDH3B	604526	244	100	100	100
IDS	300823	91	100	98	94
IDUA	252800	224	100	100	98
IMPAD1	614010	192	100	100	100
IMPDH1	146690	255	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
INPPL1	600829	195	100	100	99
ISCU	611911	91	100	100	100
ITPA	147520	208	100	98	93
IVD	607036	170	100	100	100
KHK	614058	255	100	100	100
KYNU	605197	65	99	82	71
L2HGDH	609584	73	97	96	93
LAMP2	309060	54	100	89	76
LARGE1	603590	204	100	100	100
LBR	600024	72	96	94	86
LCAT	606967	392	100	100	100
LCT	603202	230	100	100	100
LDHA	150000	81	100	98	92
LDLR	606945	268	100	100	100
LFNG	602576	221	88	86	85
LIPA	613497	85	100	100	99
LIPC	151670	151	100	92	91
LIPI	609252	49	95	87	76
LMBRD1	612625	51	89	71	61
LPIN1	605518	118	100	99	95
LPIN2	605519	125	100	100	99
LPL	609708	124	100	100	99
LRP2	600073	118	100	99	97
LRPPRC	607544	61	98	90	79
LTC4S	246530	118	100	100	98
LYST	606897	87	97	92	89
MAN1B1	604346	251	100	100	100
MAN2B1	609458	213	100	100	100
MANBA	609489	93	100	99	94
MAOA	309850	74	100	99	92
MAT1A	610550	273	100	100	100
MAT2A	601468	102	100	100	96
MCCC1	609010	111	100	96	95
MCCC2	609014	99	100	98	95
MCEE	608419	115	100	100	99
MCOLN1	605248	216	100	100	100
MFSD8	611124	82	99	93	83
MGAT2	602616	328	100	100	100
MLYCD	606761	134	100	100	99
MMAA	607481	82	100	99	95
MMAB	607568	156	100	100	100
MMACHC	609831	205	100	100	100
MMADHC	611935	56	85	85	80
MMUT	609058	101	100	95	92

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
MOCOS	613274	150	100	100	97
MOCS1	603707	198	100	100	100
MOCS2	603708	81	100	100	97
MOCS3	609277	432	100	100	100
MOGS	601336	229	100	100	100
MPDU1	604041	164	100	100	100
MPI	154550	228	100	100	100
MPV17	137960	133	100	100	99
MRPS16	609204	119	100	100	100
MRPS22	605810	72	100	98	85
MSMO1	607545	50	100	93	81
MTHFD1	172460	118	100	99	96
MTHFR	607093	191	100	100	100
MTHFS	604197	138	100	100	100
MTM1	300415	38	95	76	55
MTR	156570	119	100	100	98
MTRR	602568	95	100	100	96
MTTP	157147	89	100	100	98
MVK	251170	207	100	100	100
NAGA	104170	226	100	100	100
NAGLU	609701	182	100	100	100
NAGS	608300	311	100	100	100
NANS	605202	140	100	100	99
NAT8L	610647	265	98	91	86
NDUFA1	300078	106	100	100	100
NDUFA11	612638	243	100	100	100
NDUFA2	602137	189	100	100	100
NDUFAF1	606934	87	100	100	98
NDUFAF2	609653	77	100	99	90
NDUFAF3	612911	230	100	100	100
NDUFAF4	611776	61	90	53	38
NDUFAF5	612360	73	96	94	87
NDUFS1	157655	90	100	99	96
NDUFS2	602985	143	100	100	100
NDUFS3	603846	180	100	100	100
NDUFS4	602694	88	100	100	98
NDUFS6	603848	99	100	100	95
NDUFS7	601825	250	100	100	100
NDUFS8	602141	334	100	100	100
NDUFV1	161015	257	100	100	100
NDUFV2	600532	68	98	90	77
NEU1	608272	238	100	100	100
NFU1	608100	56	88	75	70
NHLRC1	608072	312	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NPC1	607623	138	100	100	99
NPC2	601015	119	100	100	99
NPRL2	607072	317	100	100	100
NR5A2	604453	127	100	100	97
NSDHL	300275	88	100	100	95
NT5C	191720	149	100	100	100
NT5C3A	606224	76	96	79	71
NT5E	129190	164	100	100	99
NUBPL	613621	108	100	95	88
NUS1	610463	109	93	82	70
OAT	613349	75	89	83	73
OCRL	300535	75	100	98	91
OGDH	613022	195	100	100	100
OPA1	605290	55	90	79	67
OPA3	606580	317	100	100	100
OPLAH	614243	347	100	100	100
OTC	300461	49	99	86	71
OXCT1	601424	93	100	100	95
PAH	612349	121	100	100	99
PANK2	606157	117	100	99	97
PC	608786	344	100	100	100
PCBD1	126090	127	100	100	100
PCCA	232000	71	98	93	87
PCCB	232050	159	100	98	95
PCK1	614168	214	100	100	100
PCK2	614095	245	100	100	100
PDHA1	300502	78	100	94	87
PDHB	179060	104	100	100	99
PDHX	608769	79	100	95	89
PDK1	602524	89	100	99	92
PDK2	602525	224	100	100	100
PDK3	300906	67	100	97	90
PDK4	602527	91	100	97	87
PDP1	605993	194	100	100	100
PDP2	615499	268	100	100	100
PDSS1	607429	69	100	96	82
PDSS2	610564	152	99	93	88
PDXK	179020	254	100	100	100
PEPD	613230	216	100	100	100
PEX1	602136	80	95	94	90
PEX10	602859	266	100	100	100
PEX11B	603867	105	100	99	95
PEX12	601758	131	100	100	100
PEX13	601789	113	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PEX14	601791	188	100	100	100
PEX16	603360	212	100	100	100
PEX19	600279	205	100	100	100
PEX2	170993	93	100	100	100
PEX26	608666	151	100	100	100
PEX3	603164	60	100	97	88
PEX5	600414	172	100	100	100
PEX6	601498	253	100	100	99
PEX7	601757	94	100	100	94
PFKM	610681	157	100	100	100
PGAM2	612931	286	100	100	100
PGAP1	611655	42	92	74	56
PGAP2	615187	219	100	100	100
PGAP3	611801	208	100	100	100
PGK1	311800	89	100	100	95
PGM1	171900	121	100	100	99
PGM3	172100	97	100	97	91
PHGDH	606879	199	100	100	100
PHKA1	311870	71	100	98	91
PHKA2	300798	94	100	98	92
PHKB	172490	86	98	93	87
PHKG1	172470	268	100	100	100
PHKG2	172471	307	100	100	100
PHYH	602026	99	100	97	87
PIGA	311770	62	100	99	86
PIGC	601730	139	100	100	100
PIGG	616918	146	100	99	96
PIGL	605947	159	100	100	100
PIGM	610273	151	100	100	100
PIGN	606097	50	92	79	65
PIGO	614730	253	100	100	100
PIGQ	605754	264	100	100	100
PIGT	610272	194	100	100	100
PIGV	610274	186	100	100	100
PIGW	610275	131	100	100	100
PIGY	610662	69	100	100	100
PIK3R1	171833	65	100	93	82
PIK3R5	611317	219	100	100	100
PKLR	609712	320	100	100	100
PLA2G6	603604	287	100	100	100
PLCB1	607120	80	98	94	87
PLIN1	170290	238	100	100	100
PLOD1	153454	233	100	100	100
PLOD2	601865	68	91	89	83

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PLOD3	603066	212	100	100	100
PLPBP	604436	120	100	100	99
PMM2	601785	115	100	100	99
PMPCA	613036	189	100	100	100
PNLIP	246600	96	100	100	98
PNP	164050	329	100	100	98
PNPLA2	609059	214	100	100	100
PNPLA6	603197	269	100	100	100
PNPO	603287	181	100	100	100
POFUT1	607491	199	100	100	100
POGLUT1	615618	87	100	99	91
POLG	174763	188	100	100	100
POLG2	604983	110	97	89	87
POLR3A	614258	143	100	100	99
POLR3B	614366	93	100	97	92
POMGNT1	606822	228	100	100	100
POMGNT2	614828	358	100	100	100
POMK	615247	206	100	100	100
POMT1	607423	155	100	100	100
POMT2	607439	154	100	100	97
POR	124015	296	100	100	100
PPM1K	611065	152	100	99	96
PPOX	600923	195	100	100	100
PPT1	600722	145	100	100	100
PRKAG2	602743	154	93	90	86
PRKCSH	177060	260	100	100	100
PRODH	606810	222	100	99	98
PRPS1	311850	70	100	98	92
PSAP	176801	134	100	100	99
PSAT1	610936	103	100	98	90
PSPH	172480	97	96	83	78
PTS	612719	119	100	100	100
PUS1	608109	233	100	100	100
PYCR1	179035	285	100	100	100
PYGL	613741	128	100	100	99
PYGM	608455	232	100	100	100
QDPR	612676	134	100	100	100
RARS2	611524	70	100	93	77
RBCK1	610924	200	100	100	100
RBP4	180250	281	100	100	100
RBSN	609511	172	100	100	99
RFT1	611908	134	100	95	89
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RNASEH2C	610330	261	100	100	100
RNASET2	612944	99	93	92	84
RPIA	180430	151	100	100	100
RRM2B	604712	117	100	100	98
RXYLT1	605862	75	99	91	82
SAMHD1	606754	97	100	99	93
SAR1B	607690	66	99	88	88
SARDH	604455	274	100	100	100
SC5D	602286	79	100	98	90
SCARB2	602257	79	100	97	91
SCN3A	182391	92	100	96	90
SCO1	603644	245	100	100	100
SCO2	604272	377	100	100	100
SCP2	184755	81	98	90	90
SDHA	600857	155	100	100	100
SDHAF1	612848	141	100	100	100
SDHB	185470	123	100	100	98
SDHC	602413	143	100	100	100
SDHD	602690	123	100	100	100
SEC23B	610512	125	97	97	96
SEPSECS	613009	88	99	96	90
SERAC1	614725	98	98	95	89
SGSH	605270	272	99	94	94
SHMT1	182144	177	100	100	100
SHMT2	138450	252	100	100	100
SI	609845	65	94	87	79
SLC12A3	600968	199	100	100	99
SLC16A1	600682	117	100	100	99
SLC17A5	604322	82	100	94	85
SLC19A1	600424	308	100	100	100
SLC19A2	603941	208	100	99	96
SLC19A3	606152	108	100	100	100
SLC1A1	133550	114	100	100	99
SLC1A3	600111	106	100	99	95
SLC22A5	603377	177	100	100	99
SLC23A2	603791	138	100	99	95
SLC25A1	190315	204	100	100	100
SLC25A12	603667	94	100	100	97
SLC25A13	603859	88	100	98	89
SLC25A15	603861	152	100	100	100
SLC25A19	606521	168	100	100	100
SLC25A20	613698	155	100	100	100
SLC25A22	609302	317	100	100	100
SLC25A3	600370	131	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC25A32	610815	106	100	100	98
SLC25A38	610819	181	100	100	100
SLC25A4	103220	200	100	100	100
SLC2A1	138140	354	100	100	100
SLC2A2	138160	76	100	98	94
SLC30A10	611146	209	100	100	100
SLC33A1	603690	147	98	93	85
SLC35A1	605634	69	100	99	95
SLC35A3	605632	49	93	85	76
SLC35C1	605881	329	100	100	100
SLC35D1	610804	89	92	88	76
SLC36A2	608331	135	100	98	93
SLC37A4	602671	193	100	100	100
SLC39A14	608736	159	100	100	100
SLC39A4	607059	284	100	100	100
SLC39A8	608732	91	100	93	85
SLC3A1	104614	133	99	96	89
SLC40A1	604653	105	100	98	96
SLC46A1	611672	271	100	100	100
SLC52A1	607883	285	100	100	100
SLC52A2	607882	210	100	100	100
SLC52A3	613350	271	100	100	100
SLC5A1	182380	136	100	100	98
SLC5A2	182381	258	100	100	100
SLC6A19	608893	229	100	100	100
SLC6A8	300036	134	100	100	97
SLC7A7	603593	146	100	100	100
SLC7A9	604144	154	100	100	99
SLCO1B1	604843	49	94	80	66
SLCO1B3	605495	48	94	81	67
SMPD1	607608	377	100	100	100
SOD1	147450	154	100	100	100
SPR	182125	142	100	100	100
SPTLC1	605712	98	100	99	95
SPTLC2	605713	111	100	100	99
SRD5A3	611715	138	100	100	100
SSR3	606213	97	100	98	83
SSR4	300090	139	100	100	100
ST3GAL3	606494	156	100	100	100
ST3GAL5	604402	100	100	93	92
STAR	600617	186	100	100	100
STS	300747	106	100	100	97
STT3A	601134	119	100	100	100
STT3B	608605	67	99	92	83

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SUCLA2	603921	74	100	98	88
SUCLG1	611224	90	100	100	96
SUGCT	609187	121	100	93	90
SUMF1	607939	174	100	100	100
SUOX	606887	254	100	100	100
SURF1	185620	158	100	98	95
TACO1	612958	109	100	100	100
TALDO1	602063	133	100	100	100
TANGO2	616830	253	100	100	100
TAT	613018	141	100	100	100
TAZ	300394	145	100	100	100
TBXAS1	274180	174	100	100	96
TCIRG1	604592	242	100	100	100
TCN1	189905	73	97	92	84
TCN2	613441	190	100	100	100
TDO2	191070	55	100	92	74
TECR	610057	156	100	100	100
TFR2	604720	243	100	100	100
TH	191290	266	100	100	100
TIMM8A	300356	98	100	100	100
TK2	188250	119	100	97	88
TMEM165	614726	92	100	99	93
TMEM199	616815	142	100	100	100
TMEM70	612418	103	100	98	94
TPI1	190450	190	100	100	100
TPMT	187680	61	100	95	86
TPP1	607998	223	100	100	100
TRAPPC11	614138	77	97	94	90
TREH	275360	218	100	100	99
TREX1	606609	313	100	100	100
TRIM37	605073	69	100	97	91
TRMU	610230	192	100	99	91
TRPM6	607009	192	98	97	96
TSFM	604723	182	100	99	97
TTC19	613814	101	100	98	93
TTPA	600415	93	100	100	97
TUFM	602389	218	100	100	100
TUSC3	601385	101	100	100	96
TWNK	606075	231	100	100	100
TYMP	131222	204	100	100	100
TYMS	188350	172	100	100	100
TYR	606933	147	100	99	98
TYRP1	115501	137	100	100	100
UGT1A1	191740	273	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
UMOD	191845	259	100	100	99
UMPS	613891	146	100	100	99
UPB1	606673	213	100	100	100
UQCRB	191330	98	100	100	97
UQCQRQ	612080	169	100	100	91
UROC1	613012	245	100	100	100
UROD	613521	191	100	100	100
UROS	606938	95	100	100	100
USF1	191523	171	100	100	100
VKORC1	608547	146	100	98	88
VPS13B	607817	98	99	95	89
XDH	607633	164	100	100	100
XYLT1	608124	191	100	98	95
XYLT2	608125	298	100	100	99
YARS2	610957	213	100	98	95

- 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