

WES METABOLIC DISORDERS DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AASS	137.6	99.9	99.8	238700;268700
ABAT	92.7	99.9	97.8	613163
ABCC8	124.9	100.0	99.5	600509
ABCD1	80.1	76.0	72.6	300100
ABCD2	180.0	100.0	99.8	601081
ABCD3	110.8	99.4	98.0	170995
ABCD4	135.3	99.8	97.7	614857
ABCG5	145.4	99.9	99.9	210250
ABCG8	144.9	99.0	97.1	210250;611465
ABHD12	81.7	91.7	86.0	612674
ABHD5	190.2	100.0	100.0	275630
ACACA	114.7	98.4	97.9	613933
ACAD8	132.5	100.0	100.0	611283
ACAD9	124.7	100.0	99.8	611126
ACADM	121.7	99.8	97.9	201450
ACADS	134.5	100.0	99.4	201470
ACADSB	109.9	99.8	97.9	610006
ACADVL	124.7	99.7	96.6	201475
ACAT1	114.1	99.6	97.9	203750
ACAT2	115.6	100.0	99.9	614055
ACBD5	150.7	100.0	98.4	616618
ACO2	128.7	94.1	86.3	614559
ACOX1	120.3	100.0	99.3	264470
ACOX2	106.1	100.0	99.0	601641
ACSF3	145.9	100.0	99.5	614265

ACSL4	112.9	98.3	94.2	300387
ACY1	134.7	100.0	99.7	609924
ADA	96.7	99.7	96.1	102700
ADCK5	131.2	100.0	100.0	-
ADCY5	133.1	95.9	92.5	606703
ADK	80.8	83.3	79.7	614300
ADSL	133.4	99.2	98.6	103050
AGA	159.9	100.0	99.9	208400
AGK	104.4	90.4	87.9	259900
AGL	153.8	99.8	99.5	212350;614691
AGPAT2	147.6	99.0	94.0	232400
AGPS	73.9	98.8	95.2	608594
AGXT	171.0	100.0	100.0	600121
AHCY	131.7	99.9	98.8	613752
AK1	139.4	100.0	100.0	612631
AK2	96.5	98.7	95.2	267500
AKR1C1	123.9	94.5	87.4	No OMIM phenotype
AKR1D1	95.3	99.8	98.6	235555
ALAD	97.5	97.8	92.5	612740
ALAS2	78.1	98.7	93.2	300751;300752
ALDH18A1	120.0	100.0	99.9	219150
ALDH1A3	100.0	97.6	94.6	615113
ALDH2	120.3	100.0	100.0	610251
ALDH3A2	112.0	88.8	88.4	270200
ALDH4A1	126.2	100.0	99.7	239510
ALDH5A1	93.1	92.4	83.5	271980
ALDH6A1	106.5	100.0	99.6	614105
ALDH7A1	65.7	91.1	84.5	266100
ALDOA	163.2	98.9	96.3	611881
ALDOB	127.9	98.8	95.7	229600

ALG1	44.0	53.6	46.9	608540
ALG10	295.4	100.0	99.7	613688
ALG11	133.4	96.8	96.8	613661
ALG12	149.3	100.0	99.9	607143
ALG13	81.5	97.4	90.0	300884
ALG14	206.7	100.0	99.9	612866
ALG2	90.2	100.0	100.0	607906
ALG3	90.4	100.0	99.5	601110
ALG6	111.2	98.2	93.7	603147
ALG8	107.8	96.6	95.9	608104
ALG9	103.8	99.9	99.3	608776
ALOX12B	129.4	100.0	99.7	242100
ALPL	143.9	100.0	99.4	146300;241510;241500
AMACR	151.6	100.0	100.0	214950;614307
AMN	86.5	92.5	82.9	261100
AMPD1	120.4	100.0	99.5	615511
AMPD3	119.4	100.0	99.2	612874
AMT	158.6	100.0	100.0	605899
AP1S1	105.8	99.9	99.4	609313
AP3B2	113.6	93.3	89.8	617276
APOA5	185.0	100.0	99.9	606368
APOC2	107.4	100.0	100.0	207750
APRT	90.4	100.0	100.0	614723
ARG1	134.7	92.9	92.9	207800
ARSA	120.1	100.0	99.8	250100
ARSB	111.8	98.8	91.0	253200
ASAH1	136.2	99.1	97.3	159950;228000
ASL	118.5	100.0	99.7	207900
ASNS	80.7	98.1	91.2	108370
ASPA	132.4	99.9	99.1	271900

ASS1	95.0	93.2	83.2	215700
ATIC	122.1	99.8	99.1	608688
ATP1A1	120.6	100.0	99.8	182310
ATP6AP1	102.0	98.2	93.0	300197
ATP6AP2	52.2	89.9	69.7	301045
ATP6V0A2	116.2	99.9	98.7	219200;278250
ATP6V1A	137.1	99.8	98.4	607027
ATP6V1E1	66.7	92.5	86.1	108746
ATP7A	117.5	98.7	96.0	309400
ATP7B	127.3	99.9	99.2	277900
ATP8B1	121.9	96.6	93.5	243300
AUH	108.9	99.7	99.4	250950
B3GALNT1	136.6	100.0	100.0	615021
B3GALNT2	98.9	94.3	89.8	615181
B3GALT6	53.2	77.0	73.0	615291
B3GAT3	115.0	99.4	96.6	245600
B3GLCT	100.4	99.7	98.2	261540
B4GALT1	102.1	100.0	99.3	607091
B4GALT7	124.9	99.7	96.8	130070
B4GAT1	120.2	100.0	100.0	615287
BAAT	112.4	99.5	97.5	607748
BCAT1	148.2	100.0	100.0	113520
BCAT2	150.1	100.0	100.0	113530
BCKDHA	192.2	99.8	97.9	248600
BCKDHB	125.5	99.8	95.4	248600
BCKDK	217.2	100.0	100.0	614901
BCO1	128.3	100.0	100.0	115300
BLVRA	93.4	99.8	97.8	614156
BMP2	167.5	100.0	100.0	235200
BPGM	89.2	100.0	100.0	222800

BTD	106.1	83.0	82.9	253260
C1GALT1C1	139.4	100.0	99.1	300622
CA5A	100.8	87.6	85.6	114761
CAD	139.7	99.8	98.5	114010
CANT1	145.3	100.0	100.0	251450
CAT	141.0	100.0	100.0	614097
CBLIF	117.5	100.0	99.6	609342
CBS	129.8	99.9	98.5	236200
CCDC115	98.8	95.8	90.2	613734
CD320	110.1	100.0	100.0	606475
CEL	166.7	90.8	89.2	609812
CERKL	115.1	99.3	96.5	608380
CERS3	93.4	99.7	98.0	615276
CFTR	124.9	99.5	97.9	167800;277180;219700;211400;614122
CHIT1	105.2	99.9	98.2	600031
CHKB	111.3	100.0	99.6	602541
CHST14	149.1	99.9	98.8	601776
CHST3	127.4	100.0	99.9	245600;143095
CHST6	216.4	100.0	100.0	217800
CHSY1	122.1	97.8	96.3	605282
CLCN7	143.3	99.4	97.8	618541
CLN3	128.6	92.5	92.4	204200
CLN5	106.5	69.0	66.3	256731
CLN6	136.4	99.9	98.9	601780
CLN8	152.5	83.5	83.5	600143
CLPB	121.2	94.9	94.0	616271
CMAS	97.2	99.3	96.7	603316
COG1	109.3	100.0	99.9	611209
COG2	117.5	99.8	98.9	606974
COG4	99.4	100.0	100.0	613489

COG5	136.7	99.1	96.8	613612
COG6	97.2	98.5	93.1	614576
COG7	109.9	100.0	99.4	608779
COG8	125.4	98.6	95.3	611182
COMT	156.5	100.0	100.0	167870;181500
COQ2	83.7	97.6	96.7	609825
COQ4	92.0	91.0	89.7	616276
COQ5	151.1	100.0	100.0	616359
COQ6	118.1	99.9	98.5	614647
COQ7	121.1	100.0	99.6	601683
COQ8A	166.1	100.0	99.6	606980
COQ8B	102.1	100.0	99.2	615567
COQ9	69.5	100.0	98.7	612837
CP	108.0	92.6	85.2	604290
CPOX	128.4	99.8	97.2	121300
CPS1	138.3	100.0	100.0	237300;265380
CPT1A	141.6	99.8	97.6	255120
CPT2	148.5	98.2	97.4	255110;608836;614212;600649
CRAT	126.7	100.0	99.9	600184
CRPPA	105.9	98.4	94.7	614643;616052
CTH	150.4	100.0	99.9	219500
CTNS	117.6	100.0	99.3	219800;219900;219750
CTSA	139.9	100.0	99.6	256540
CTSC	123.6	100.0	100.0	170650;245010;245000
CTSD	174.4	98.4	95.0	610127
CTSK	90.8	100.0	99.2	265800
CUBN	107.3	99.2	97.1	261100
CYB561	152.3	92.8	92.7	600019
CYB5R3	161.6	99.1	98.1	250800
CYP11A1	119.9	99.2	94.5	613743

CYP11B1	155.9	100.0	99.9	202010;103900
CYP11B2	154.9	100.0	99.9	610600;203400
CYP17A1	109.0	99.9	98.5	202110
CYP19A1	137.2	98.3	95.7	613546;139300
CYP1B1	96.4	100.0	100.0	231300;604229
CYP21A2	101.2	97.4	91.1	201910
CYP27A1	165.1	99.7	98.1	213700
CYP27B1	129.3	100.0	99.8	264700
CYP2R1	137.6	99.5	96.0	600081
CYP2U1	125.9	95.3	92.0	615030
CYP7B1	111.4	98.1	92.7	270800;613812
D2HGDH	142.4	99.7	98.2	600721
DAO	109.8	100.0	99.9	181500
DBH	171.0	100.0	100.0	223360
DBT	112.5	99.1	96.1	248600
DCXR	177.3	99.1	94.4	260800
DDC	101.9	99.2	95.0	608643
DDHD1	142.3	98.5	96.5	609340
DDOST	113.4	100.0	99.8	614507
DEGS1	139.6	100.0	100.0	615843
DGAT1	155.6	91.8	87.6	615863
DGKE	123.8	99.7	98.5	615008
DGUOK	112.4	99.9	98.8	251880
DHCR24	154.7	97.7	97.7	602398
DHCR7	147.5	100.0	100.0	270400
DHDDS	80.8	99.4	95.6	608172
DHFR	43.9	88.9	76.3	613839
DHODH	106.1	100.0	99.9	263750
DLD	112.4	99.9	99.7	246900
DMGDH	144.8	99.9	99.7	605850

DNAJC12	155.7	87.4	87.3	606060
DNAJC19	105.0	99.3	92.2	610198
DNM1L	118.8	99.6	98.3	614388
DNM2	133.5	98.6	93.9	606482;160150
DNMT1	117.3	99.2	98.8	614116
DNMT3B	123.2	100.0	99.9	242860
DOLK	175.4	100.0	100.0	610768
DPAGT1	79.4	100.0	99.8	608093;614750
DPM1	129.7	97.4	90.9	608799
DPM2	87.3	100.0	97.7	615042
DPM3	229.1	100.0	100.0	612937
DPYD	146.1	99.5	96.5	274270
DPYS	118.4	100.0	100.0	222748
DTYMK	108.6	100.0	100.0	188345
EBP	67.1	99.5	94.3	302960
ECHS1	97.3	100.0	99.4	616277
ELOVL1	87.5	99.6	96.5	611813
ELOVL4	104.8	99.7	98.9	600110;614457
ENO3	173.9	100.0	100.0	612932
EOGT	108.0	79.3	77.8	614789
EPHX1	123.9	99.8	97.8	189800;607748
EPHX2	95.8	99.5	96.5	143890
ETFA	139.1	99.8	99.6	231680
ETFB	125.0	100.0	99.9	231680
ETFDH	123.6	99.8	99.4	231680
ETHE1	85.4	99.3	93.3	602473
EXT1	89.6	99.6	97.1	133700;215300
EXT2	124.9	99.9	99.0	133701
EYA1	120.9	99.9	99.5	601653
FA2H	86.5	92.4	82.6	612319

FAH	122.1	100.0	99.5	276700
FAR1	77.6	97.4	94.0	616107
FBP1	89.9	93.6	91.3	229700
FCSK	100.8	98.0	96.1	608675
FDFT1	133.8	98.5	96.7	184420
FECH	101.9	99.9	99.8	177000
FH	128.8	93.2	87.2	606812
FKRP	140.6	100.0	100.0	613153;606612;606596;607155
FKTN	108.4	99.8	95.2	613152;611615;611588;253800
FLAD1	181.4	100.0	99.7	610595
FMO3	141.4	99.9	99.7	602079
FOLR1	103.2	100.0	99.9	613068
FTCD	106.4	97.7	93.2	229100
FUCA1	126.0	100.0	100.0	230000
FUT2	150.7	100.0	100.0	612542
FUT6	146.3	100.0	100.0	613852
FUT8	127.5	99.8	98.9	602589
G6PC	163.3	100.0	100.0	232200
G6PC3	116.4	100.0	99.9	612541
G6PD	114.3	99.1	97.4	134700;611162
GAA	177.6	100.0	99.9	232300
GAD1	117.4	100.0	99.3	603513
GALC	98.7	99.7	97.6	245200
GALE	149.2	100.0	100.0	230350
GALK1	156.6	100.0	99.2	230200
GALM	93.5	100.0	99.5	137030
GALNS	110.5	100.0	99.3	253000
GALNT2	117.7	99.8	97.1	618885
GALNT3	140.2	99.8	98.7	601756
GALT	160.1	100.0	99.6	230400

GAMT	124.7	95.0	82.7	612736
GANAB	109.2	99.8	97.8	104160
GATM	140.6	100.0	100.0	612718
GBA	179.6	100.0	100.0	230900;127750;231005;231000;168600;608013;230800
GBA2	151.2	100.0	99.5	614409
GBE1	172.1	99.9	99.7	263570;232500
GCDH	157.8	100.0	99.2	231670
GCH1	78.0	99.9	97.3	233910;128230
GCK	130.0	95.4	95.4	125853;606176;125851;602485
GCLC	143.7	99.4	97.1	608446;230450
GCLM	94.4	99.5	95.4	608446
GCSH	27.8	75.7	64.4	605899
GFPT1	164.5	99.9	99.4	610542;608931
GGPS1	152.9	99.8	99.8	606982
GK	44.9	84.2	61.8	307030
GLA	72.8	91.0	85.9	301500
GLB1	77.0	99.2	92.8	230650;253010;230600;230500
GLDC	57.7	88.9	77.8	605899
GLRA1	94.7	100.0	99.8	149400
GLRX5	118.9	97.2	89.6	205950
GLS	79.5	96.9	88.5	138280
GLUD1	64.6	96.4	84.4	606762
GLUL	63.1	73.0	69.0	610015
GLYCTK	163.6	98.7	97.3	220120
GM2A	119.1	100.0	100.0	272750
GMPPA	163.9	100.0	100.0	615495
GMPPB	257.0	100.0	100.0	615352;615351;615350
GMPS	111.9	98.2	94.5	601626
GNE	118.9	100.0	99.5	269921;600737;605820
GNMT	128.9	100.0	100.0	606664

GNPAT	124.5	99.5	95.6	222765
GNPTAB	148.1	99.9	99.7	252500;252600
GNPTG	144.9	99.8	96.6	252605
GNS	95.6	99.2	94.6	252940
GOT1	108.1	100.0	98.6	614419
GOT2	74.6	94.6	87.0	138150
GPD1	94.0	100.0	99.9	614480
GPD1L	124.4	100.0	98.8	611777
GPHN	153.4	99.9	99.1	252150
GPI	148.8	100.0	99.3	613470
GPIHBP1	162.7	100.0	99.9	612757
GPT2	123.8	99.4	95.3	138210;616281
GPX1	57.7	97.4	88.7	614164
GRHPR	96.8	83.3	79.2	260000
GSS	96.3	96.5	96.3	231900;266130
GUSB	108.2	92.5	90.1	253220
GYG1	134.7	99.6	97.4	613507
GYS1	119.5	100.0	98.0	611556
GYS2	131.3	99.9	99.4	240600
H6PD	192.8	99.0	99.0	604931
HADH	109.8	99.2	97.7	231530;609975
HADHA	70.8	95.5	88.3	609015;609016
HADHB	73.3	97.7	87.0	609015
HAGH	142.3	100.0	100.0	614033
HEXA	99.6	93.8	93.1	272800
HEXB	174.4	99.4	96.6	268800
HFE	101.9	99.9	97.8	235200
HGD	106.1	100.0	99.7	203500
HGSNAT	109.4	86.4	86.2	252930
HIBADH	110.8	93.8	91.3	608475

HIBCH	70.8	98.2	84.5	250620
HK1	120.9	100.0	99.9	235700
HLCS	153.7	100.0	100.0	253270
HMBS	93.1	100.0	98.4	176000
HMGCL	108.0	100.0	99.4	246450
HMGCS2	106.3	100.0	99.7	605911
HMOX1	142.5	97.7	90.1	614034;606963
HNF1A	183.8	100.0	99.8	142410
HNF4A	133.8	99.9	98.6	600281
HOGA1	134.9	99.5	95.5	613616
HPD	158.2	100.0	99.8	140350;276710
HPDL	208.5	100.0	100.0	619026;619027
HPRT1	67.0	98.6	90.6	300323;300322
HS6ST1	59.3	93.6	86.7	614880
HSD11B1	114.9	100.0	99.7	614662
HSD11B2	172.2	87.6	83.8	218030
HSD17B10	103.8	99.9	98.3	300438;300705;300220
HSD17B3	112.2	97.8	97.8	264300
HSD17B4	114.3	95.3	92.8	261515;233400
HSD3B2	128.3	100.0	99.7	201810
HSD3B7	148.0	98.9	95.0	607765
HTRA2	120.2	100.0	99.6	606441
HYAL1	107.8	100.0	100.0	601492
IDH2	108.9	99.8	97.4	613657
IDH3B	127.4	95.4	95.4	612572
IDI1	59.5	99.0	96.4	604055
IDS	95.1	99.6	95.3	309900
IDUA	139.0	94.6	87.4	607015;607014;607016
IMPAD1	147.0	100.0	99.9	614078
IMPDH1	56.2	89.0	81.7	180105;613837

INPP5E	113.1	96.9	93.2	610156
INPPL1	133.9	98.6	94.4	258480
INSR	123.8	97.3	93.0	147670
IREB2	137.2	99.9	99.8	147582
ITCH	115.4	91.5	90.8	606409
ITPA	126.4	100.0	100.0	147520
IVD	90.4	100.0	99.9	243500
KCNA2	132.6	100.0	99.6	616366
KCNJ11	174.3	100.0	100.0	600937
KMT2A	129.6	100.0	99.7	605130
KMT2D	133.9	99.9	99.0	147920
L2HGDH	123.6	98.9	96.4	236792
LAMP2	88.2	99.3	96.0	300257
LARGE1	110.3	100.0	99.7	613154;608840
LCAT	124.7	98.8	93.3	136120;245900
LCT	118.4	99.6	97.4	223000
LDHA	69.1	94.4	89.3	612933
LDHB	92.6	90.5	77.9	614128
LFNG	113.2	88.6	86.5	609813
LIAS	130.5	99.8	98.9	607031
LIPA	105.7	96.9	94.6	278000
LIPC	98.8	100.0	99.4	614025;125853;612797
LIPT1	156.1	99.7	99.6	610284
LIPT2	68.5	98.4	82.4	617659
LMBRD1	93.8	94.1	89.1	277380
LMF1	143.0	100.0	99.7	611761
LPIN1	123.7	99.4	97.2	268200
LPIN2	105.3	99.9	99.7	609628
LPL	128.2	100.0	100.0	144250;238600
LRAT	288.9	100.0	100.0	268000;613341

LTC4S	69.9	76.4	69.0	614037
LYST	140.4	99.4	97.8	214500
MAN1B1	126.7	100.0	99.7	614202
MAN2B1	140.0	99.6	97.4	248500
MAN2B2	156.3	99.9	99.2	No OMIM phenotype
MANBA	103.5	87.1	84.9	248510
MAOA	101.4	100.0	99.8	300615
MAT1A	149.8	99.9	98.5	250850
MBOAT7	112.6	100.0	99.3	617188
MCCC1	149.2	99.9	98.7	210200
MCCC2	117.3	99.9	99.1	210210
MCEE	131.3	100.0	100.0	251120
MCOLN1	159.2	99.8	98.8	252650
MDH1	100.1	99.7	99.1	154200
MFSD2A	123.0	99.5	97.3	614397
MFSD8	121.3	99.6	99.4	610951
MGAT2	125.7	100.0	99.9	212066
MINPP1	167.9	99.7	99.3	188470
MLYCD	85.3	96.8	92.5	248360
MMAA	169.4	100.0	100.0	251100
MMAB	109.1	100.0	99.9	251110
MMACHC	198.3	100.0	100.0	277400
MMADHC	90.8	91.6	81.3	277410
MMUT	142.3	99.7	98.2	251000
MOCOS	157.7	99.9	97.8	613274
MOCS1	92.4	98.9	95.5	252150
MOCS2	143.2	99.4	99.4	252150
MOGS	149.2	100.0	99.9	606056
MPDU1	105.1	100.0	99.2	609180
MPI	115.0	100.0	99.5	602579

MRPL44	110.3	99.5	97.4	615395
MRPS36	60.4	94.0	75.2	611996
MSMO1	54.6	93.1	86.8	607545
MTHFD1	121.9	99.9	98.4	601634
MTHFR	111.9	97.3	95.9	181500;236250;188050;601634
MTM1	78.0	98.7	92.0	310400
MTMR2	105.7	99.5	98.4	601382
MTR	135.6	100.0	99.9	250940;601634
MTRR	129.7	99.8	98.4	236270
MVK	117.9	91.4	90.5	175900;260920;610377
NADK2	171.6	99.7	99.3	615787
NAGA	149.9	100.0	100.0	609242;609241
NAGLU	119.2	93.8	91.7	252920
NAGS	91.1	99.9	97.9	237310
NANS	98.3	100.0	99.9	605202
NAXD	131.1	100.0	99.9	618321
NAXE	80.6	100.0	98.6	608862
NBAS	141.0	99.9	99.3	608025
NEU1	144.9	99.3	96.1	256550
NGLY1	142.3	99.8	99.7	610661
NMNAT1	110.5	100.0	99.2	608553
NNT	118.6	96.4	96.0	614736
NPC1	114.4	99.9	99.0	257220
NPC2	136.0	100.0	99.2	607625
NPL	116.2	100.0	99.3	611412
NSD1	137.5	100.0	99.8	130650;601626;117550
NSDHL	132.0	99.8	96.3	308050;300831
NT5C3A	64.8	94.6	82.2	266120
NT5E	143.6	100.0	100.0	211800
NUS1	57.9	56.5	42.0	610463

OAT	72.2	82.0	73.0	258870
OCRL	107.2	99.4	97.6	300555;309000
OGDH	165.6	100.0	99.8	613022
OPA3	134.8	100.0	99.5	258501
OPLAH	145.2	100.0	99.8	260005
OTC	120.4	100.0	99.9	311250
OXCT1	129.8	99.4	97.6	245050
PAH	130.6	100.0	100.0	261600
PANK2	159.3	100.0	99.7	234200;607236
PC	158.4	99.7	98.0	266150
PCBD1	110.6	100.0	99.8	264070
PCCA	103.7	98.9	93.4	606054
PCCB	107.3	96.7	95.4	606054
PCK1	122.8	100.0	100.0	614168
PCK2	160.3	100.0	100.0	614095
PCYT1A	102.5	99.2	95.7	123695;608940
PCYT2	144.6	100.0	98.3	602679
PDSS1	110.5	95.2	87.8	607429
PDSS2	112.7	98.4	94.3	610564
PEPD	103.8	100.0	99.4	170100
PEX1	135.8	99.8	99.4	601539;214100
PEX10	105.9	98.8	90.6	614871;614870
PEX11B	89.6	100.0	98.3	614920
PEX12	132.6	100.0	100.0	614859
PEX13	168.6	100.0	100.0	614885;614883
PEX14	139.1	95.8	89.4	614887
PEX16	167.1	97.1	93.9	614876;614877
PEX19	87.0	99.0	94.4	614886
PEX2	133.2	100.0	100.0	614866;614867
PEX26	86.7	100.0	99.8	614872;614873

PEX3	98.2	99.4	99.2	614882
PEX5	110.1	99.9	98.8	202370;214110
PEX6	109.2	96.4	88.0	614862;614863
PEX7	113.8	88.0	81.0	215100;614879
PFKM	114.7	100.0	99.7	232800
PGAM2	168.4	100.0	100.0	261670
PGAP1	106.2	98.7	94.6	611655
PGAP2	138.8	100.0	99.9	614207
PGAP3	66.7	62.6	58.1	611801
PGK1	43.3	90.3	73.2	300653
PGM1	124.5	94.2	94.1	614921;612934
PGM3	147.5	99.9	99.7	172100
PHGDH	111.5	99.9	98.2	601815
PHKA1	100.8	97.8	93.4	300559
PHKA2	100.1	100.0	99.1	306000
PHKB	125.6	99.7	99.1	172490
PHKG1	120.3	99.8	97.5	172470
PHKG2	167.2	100.0	99.9	172471
PHYH	71.1	100.0	98.9	266500
PI4K2A	96.4	93.4	87.6	No OMIM phenotype
PIGA	73.9	91.6	82.5	300818;300868
PIGB	107.3	99.5	97.3	604122
PIGC	84.9	96.0	86.2	601730
PIGL	123.6	100.0	99.6	280000
PIGM	128.8	100.0	100.0	610293
PIGN	96.7	93.1	89.6	614080
PIGO	149.1	100.0	99.8	614749
PIGP	76.2	95.6	85.5	605938
PIGQ	143.1	93.4	91.6	605754
PIGT	168.8	98.1	98.0	610272

PIGV	122.2	100.0	100.0	239300
PIGW	137.3	100.0	99.7	610275
PIGY	87.8	100.0	100.0	610662
PIK3CA	128.2	97.7	97.3	114480;182000;603387;114550;167000;602501;612918;211980;114500;615108;613659;162900
PIK3R1	123.8	99.7	98.4	615214
PIK3R2	108.2	90.9	89.1	603387
PIK3R5	125.9	100.0	99.9	615217
PIKFYVE	143.0	99.9	99.3	121850
PIP5K1C	142.3	99.2	96.7	611369
PKLR	174.3	99.9	98.0	102900;266200
PLA2G5	120.0	100.0	100.0	228980
PLA2G6	111.6	92.1	90.7	610217;256600;612953
PLA2G7	127.3	99.8	99.3	614278;600807;147050
PLCB1	136.7	99.9	99.4	613722
PLCB4	99.9	99.8	98.7	614669
PLCD1	121.7	99.9	97.3	151600
PLCE1	126.1	99.8	98.9	610725
PLCG2	108.6	100.0	99.3	614878;614468
PLIN1	87.1	99.6	95.2	613877
PLOD1	140.5	100.0	98.2	225400
PLOD2	127.9	99.2	98.1	609220
PLOD3	107.9	100.0	98.7	612394
PLPBP	90.1	95.1	88.9	604436
PMM2	127.8	99.8	99.8	212065
PNLIP	147.4	99.9	99.6	614338
PNMT	96.2	99.7	96.8	145500
PNP	109.8	99.8	98.7	613179
PNPLA2	137.8	99.8	96.1	610717
PNPLA6	144.9	100.0	99.8	612020
PNPO	73.2	99.9	97.1	610090

POFUT1	130.4	100.0	98.9	607491
POGLUT1	106.6	99.8	95.9	615618
POLR3A	115.6	99.9	99.0	607694
POLR3B	131.0	99.7	97.6	607694;614381
POMGNT1	116.4	100.0	99.8	613151;253280;613157
POMGNT2	182.2	100.0	100.0	614830
POMK	152.4	100.0	100.0	615249;616094
POMT1	131.1	99.5	97.3	609308;236670;613155
POMT2	98.9	99.8	97.3	613156;236670;613158;613150
PPCS	130.5	100.0	99.1	609853
PPM1K	146.1	100.0	100.0	615135
PPOX	97.1	99.3	95.7	176200
PPT1	137.3	90.3	89.9	256730
PRKAG2	128.8	99.4	96.1	602743
PRKCSH	159.9	99.5	94.1	177060
PRODH	81.3	84.0	80.2	600850;239500
PRPS1	103.4	86.4	86.3	304500;301835;311070;300661
PSAP	99.1	100.0	99.6	611722;610539;249900;611721
PSAT1	43.1	92.0	75.1	610992
PSPH	139.8	100.0	100.0	614023
PTEN	144.3	99.5	97.2	176807;155600;607174;188470;158350;608089;153480;613028;275355;605309;276950
PTGIS	117.9	99.0	95.1	145500
PTPN11	81.2	97.7	87.6	163950;156250;607785;151100
PTS	104.8	99.5	99.0	261640
PUS3	157.8	100.0	100.0	616283
PYCR1	95.6	100.0	98.2	612940;614438
PYCR2	127.2	100.0	99.3	616420
PYGL	126.4	100.0	100.0	232700
PYGM	128.2	100.0	100.0	232600
QDPR	97.7	100.0	98.9	261630

RBCK1	102.4	99.9	98.3	610924;615895
RDH12	82.4	99.3	95.4	612712
RDH5	178.1	100.0	100.0	136880
RFT1	106.1	99.7	98.4	612015
RINT1	159.1	99.6	97.6	610089
RPE65	125.6	99.9	98.7	204100;613794
RPIA	99.6	99.1	96.1	608611
RPN2	119.6	100.0	100.0	180490
RXYLT1	145.6	99.2	95.9	615041
SARDH	132.7	93.4	91.6	268900
SAT1	127.4	99.7	96.5	308800
SC5D	178.6	99.9	99.1	607330
SCARB2	108.9	99.9	99.4	254900
SCP2	111.6	99.9	97.9	613724
SCYL1	152.8	100.0	99.9	607982
SEC23B	134.5	99.9	99.1	610512
SELENBP1	128.4	100.0	99.7	604188
SEPSECS	154.7	99.9	99.6	613811
SERAC1	108.9	99.6	99.5	614739
SGSH	143.6	94.8	94.1	252900
SI	129.5	99.0	95.9	222900
SLC10A7	107.8	99.5	98.1	611459
SLC12A1	139.8	96.2	96.0	600839
SLC13A3	95.6	99.8	97.9	606411
SLC16A1	124.8	100.0	98.6	245340
SLC17A5	116.5	99.6	96.2	604369;269920
SLC18A2	114.4	99.9	99.6	193001
SLC22A12	119.6	100.0	99.8	607096
SLC22A5	125.6	100.0	99.6	212140
SLC25A1	91.4	96.9	89.8	615182

SLC25A13	110.8	100.0	99.4	603471
SLC25A15	161.2	99.3	96.6	238970
SLC25A19	84.3	99.9	98.0	607196
SLC25A20	85.6	100.0	98.9	212138
SLC25A21	121.8	100.0	99.6	607571
SLC25A32	123.9	100.0	100.0	610815
SLC25A38	98.1	97.4	93.3	205950
SLC25A42	133.3	97.1	94.3	610823
SLC28A1	131.5	100.0	98.3	606207
SLC2A1	164.2	92.8	92.7	606777
SLC2A2	148.2	100.0	99.8	227810
SLC2A9	114.9	99.3	95.0	606142
SLC30A10	162.7	100.0	100.0	613280
SLC33A1	120.6	99.8	98.5	612539;614482
SLC35A1	124.0	99.7	99.3	603585
SLC35A2	110.5	99.6	97.7	314375
SLC35A3	72.7	80.4	78.8	605632
SLC35C1	178.7	100.0	99.4	266265
SLC35D1	114.8	99.6	97.6	610804
SLC37A4	118.5	99.8	97.6	232220
SLC39A14	102.8	100.0	99.0	608736
SLC39A4	109.1	99.3	96.3	201100
SLC39A8	126.5	100.0	99.7	608732
SLC3A1	148.6	100.0	99.7	220100
SLC44A1	153.0	98.2	98.1	606105
SLC46A1	103.5	100.0	98.5	229050
SLC52A1	210.3	100.0	100.0	615026
SLC52A2	171.9	100.0	100.0	614707
SLC52A3	127.1	100.0	100.0	211530
SLC5A1	119.7	100.0	99.8	606824

SLC5A2	136.4	100.0	100.0	233100
SLC6A19	135.9	100.0	100.0	608893
SLC6A5	123.8	100.0	99.9	614618
SLC6A8	54.5	94.8	83.0	300352
SLC6A9	155.6	100.0	99.6	617301
SLC7A7	114.4	100.0	99.9	222700
SLC7A9	132.6	100.0	99.4	220100
SLCO1B1	57.4	98.3	92.0	237450
SLCO1B3	53.3	98.1	88.4	237450
SMPD1	154.8	100.0	99.9	257200;607616
SMS	66.0	87.9	72.1	309583
SNX14	81.0	98.9	93.6	616354
SOD1	123.9	100.0	100.0	618598;105400
SOD2	195.5	100.0	100.0	147460
SPR	153.3	100.0	99.4	612716
SPTLC1	107.4	98.7	93.7	162400
SPTLC2	141.9	100.0	100.0	613640
SQOR	89.5	100.0	98.0	617658
SRD5A2	72.2	100.0	98.8	264600
SRD5A3	122.0	100.0	99.1	612713;612379
SSR4	114.0	100.0	99.5	300934
ST3GAL3	95.7	68.8	68.2	611090;615006
ST3GAL5	94.7	85.9	84.0	609056
STAR	135.7	100.0	99.9	201710
STS	78.3	96.8	93.8	308100
STT3A	135.2	100.0	100.0	601134
STT3B	127.7	99.7	99.4	608605
STX5	71.4	95.0	89.3	603189
SUCLA2	55.1	88.8	79.4	612073
SUCLG1	102.4	100.0	99.7	245400

SUCLG2	55.7	91.7	79.1	603922
SUGCT	140.2	99.6	97.6	231690;277410
SUMF1	91.4	98.3	92.5	272200
SUOX	179.3	100.0	100.0	272300
TALDO1	143.9	100.0	98.0	606003
TANGO2	127.0	100.0	99.3	616830
TAT	113.5	100.0	100.0	276600
TAZ	104.7	99.3	93.7	302060
TBXAS1	131.9	100.0	100.0	614158;231095
TCIRG1	118.4	98.5	93.4	259700
TCN2	165.7	100.0	100.0	275350
TECR	131.9	100.0	98.5	614020
TH	93.5	99.8	98.0	605407
TIMM50	126.6	98.4	95.0	607381
TK2	104.4	99.0	96.0	609560
TKFC	142.5	100.0	99.8	615844
TKT	113.2	98.6	96.8	606781
TMEM106B	126.9	99.3	98.3	617964
TMEM165	123.8	100.0	100.0	614727
TMEM199	108.1	100.0	100.0	616815
TMEM70	116.7	98.4	94.6	612418
TMLHE	91.2	98.6	94.1	300872
TPI1	117.0	99.8	98.0	615512
TPK1	99.5	99.5	97.2	606370
TPMT	36.6	98.4	82.0	610460
TPP1	130.4	100.0	100.0	204500
TRAK1	149.2	93.3	93.1	608112
TRAPPC11	133.5	99.7	98.7	614138
TRAPPC2L	186.5	100.0	100.0	610970
TREH	139.4	97.7	93.0	612119

TUSC3	171.7	100.0	99.7	611093
TYMP	111.2	100.0	99.4	603041
TYMS	124.5	100.0	99.7	188350
TYR	147.1	100.0	100.0	203100;103470;601800;606952
TYRP1	164.9	100.0	99.9	612271;203290
UGT1A1	182.3	100.0	100.0	237900;143500;601816;218800;606785
UMPS	145.5	99.9	98.7	258900
UPB1	135.4	100.0	100.0	613161
UROC1	151.8	100.0	99.9	276880
UROD	134.9	98.5	95.5	176100
UROS	100.1	100.0	99.9	263700
VMA21	82.3	99.0	92.6	300913
VPS13B	139.7	99.4	97.8	607817
VPS33A	97.1	91.9	89.9	617303
XDH	94.7	100.0	99.8	278300
XYLT1	134.5	97.8	91.1	608124
XYLT2	158.7	99.9	97.1	608125
ZBTB11	174.9	99.9	99.3	No OMIM phenotype

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

Median Coverage describes the average number of reads seen across 50 exomes.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with no value for coverage are non protein coding genes.

Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.