

# WES MITOCHONDRIAL DISORDERS DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
AARS2	131.7	100.0	99.4	612035
ABAT	92.7	99.9	97.8	613163
ACAD9	124.7	100.0	99.8	611103
ACO2	128.7	94.1	86.3	100850
ACTA1	83.4	98.2	89.5	102610
ADAMTS10	125.9	100.0	99.9	608990
ADCK2	162.8	100.0	99.7	-
ADPRS	141.6	100.0	99.9	610624
AFG3L2	98.3	94.6	86.3	604581
AGK	104.4	90.4	87.9	610345
AIFM1	103.9	99.9	97.8	300169
ALDH1B1	191.3	100.0	100.0	100670
ALKBH1	99.3	100.0	99.7	605345
ANO10	116.3	99.2	96.6	613726
APOO	57.6	81.1	71.1	No OMIM phenotype
APTX	98.5	94.1	90.6	606350
ARL2	136.0	100.0	99.7	601175
ARNT2	139.0	100.0	100.0	606036
ATAD1	62.6	99.1	91.6	614452
ATAD3A	91.9	91.4	86.7	612316
ATAD3B	88.2	91.1	83.2	612317
ATP13A2	141.0	100.0	99.6	610513
ATP5F1A	68.0	92.2	83.0	164360
ATP5F1B	110.0	99.8	97.7	102910
ATP5F1C	75.0	95.5	89.3	108729

ATP5F1D	86.1	97.4	91.8	603150
ATP5F1E	148.5	100.0	100.0	606153
ATP5IF1	152.6	100.0	100.0	614981
ATP5MC1	91.7	100.0	99.6	603192
ATP5MC2	70.0	99.3	92.4	603193
ATP5MC3	120.8	100.0	100.0	602736
ATP5MD	17.8	83.8	35.7	615204
ATP5ME	77.7	100.0	100.0	601519
ATP5MF	117.2	99.6	94.8	-
ATP5MG	115.6	100.0	100.0	617473
ATP5MGL	177.9	100.0	100.0	613213
ATP5PB	57.4	97.0	83.3	603270
ATP5PD	82.7	89.2	67.8	618121
ATP5PF	63.8	99.9	92.4	603152
ATP5PO	89.5	100.0	98.1	600828
ATPAF1	70.8	83.7	71.4	608917
ATPAF2	99.7	100.0	99.9	608918
BCAP31	67.6	92.1	79.1	300475
BCS1L	143.8	100.0	100.0	603647
BOLA1	98.8	100.0	99.9	613181
BOLA2	108.9	100.0	100.0	613182
BOLA3	46.4	99.0	86.7	613183
C12orf65	78.9	99.0	94.5	613541
C19orf12	122.8	100.0	99.8	614297
C1QBP	66.9	84.3	70.6	601269
CA5A	100.8	87.6	85.6	114761
CARS2	122.9	100.0	100.0	612800
CEP89	137.3	95.8	94.5	615470
CFAP58	88.5	99.7	97.1	No OMIM phenotype
CHCHD10	27.7	57.8	42.0	615903

CHCHD2	70.5	93.7	78.7	616710
CHKB	111.3	100.0	99.6	612395
CISD2	112.9	83.4	83.4	611507
CLPB	121.2	94.9	94.0	616271
CLPP	146.0	100.0	99.5	601119
COA1	89.3	100.0	100.0	614769
COA3	124.6	100.0	100.0	614775
COA5	66.1	94.4	83.0	613920
COA6	96.8	99.6	96.3	614772
COA7	115.6	100.0	100.0	615623
COA8	71.5	81.9	80.8	616003
COASY	173.4	100.0	100.0	609855
COQ2	83.7	97.6	96.7	609825
COQ4	92.0	91.0	89.7	612898
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
COX10	194.2	100.0	99.9	602125
COX14	100.4	100.0	100.0	614478
COX15	91.4	99.9	97.8	603646
COX16	76.1	99.3	96.9	618064
COX20	62.9	95.7	82.4	614698
COX411	107.2	100.0	100.0	123864
COX412	125.7	100.0	99.9	607976
COX5A	24.1	66.0	36.0	603773
COX5B	139.4	100.0	100.0	123866
COX6A1	158.2	100.0	99.9	602072

COX6A2	52.2	99.8	94.0	602009
COX6B1	169.3	100.0	100.0	124089
COX6B2	81.4	100.0	99.9	220110
COX6C	100.4	99.5	94.3	124090
COX7A1	123.8	100.0	99.9	123995
COX7A2	95.6	100.0	98.8	123996
COX7B	43.8	69.1	35.9	603792
COX7B2	130.9	100.0	100.0	609811
COX7C	33.6	98.4	78.9	603774
COX8A	101.3	100.0	100.0	123870
COX8C	130.5	100.0	99.9	220110
CP	108.0	92.6	85.2	117700
CRAT	126.7	100.0	99.9	600184
CTBP1	108.5	94.3	86.9	602618
CYC1	147.7	98.3	89.5	123980
CYCS	68.9	99.4	96.9	123970
DARS2	120.7	94.8	93.8	610956
DCAF17	92.0	98.5	93.4	612515
DDHD1	142.3	98.5	96.5	614603
DES	115.4	100.0	99.6	125660
DGUOK	112.4	99.9	98.8	601465
DHTKD1	123.4	99.8	98.8	614984
DLAT	98.4	99.8	99.3	608770
DLD	112.4	99.9	99.7	238331
DLST	92.4	95.7	87.7	126063
DMAC1	59.4	100.0	99.9	617261
DMAC2	132.6	98.3	98.3	617262
DMAC2L	104.8	99.8	99.8	603152
DNA2	132.9	99.6	96.9	601810
DNAJA3	126.3	98.5	96.1	608382

DNAJC19	105.0	99.3	92.2	608977
DNAJC3	121.4	99.7	99.7	601184
DNAJC30	149.9	100.0	100.0	618202
DNM1L	118.8	99.6	98.3	603850
EARS2	102.2	99.8	98.0	612799
ECHS1	97.3	100.0	99.4	602292
ECSIT	149.0	100.0	100.0	608388
EHHADH	146.4	100.0	100.0	607037
ELAC2	109.3	100.0	99.2	605367
ERAL1	164.8	100.0	99.6	607435
ETFDH	123.6	99.8	99.4	231675
ETHE1	85.4	99.3	93.3	608451
FA2H	86.5	92.4	82.6	611026
FARS2	180.1	100.0	100.0	614946
FARSB	81.0	98.0	92.9	609690
FASTKD2	119.2	99.6	98.6	612322
FBXL4	166.7	100.0	100.0	605654
FDX2	124.7	100.0	100.0	614585
FDXR	131.6	100.0	98.6	103270
FH	128.8	93.2	87.2	606945
FOXRED1	121.6	100.0	99.6	613622
FTL	134.8	98.6	88.5	134790
FXN	66.0	98.3	84.7	606829
GARS1	131.4	99.9	99.4	600287
GATB	104.7	100.0	99.0	603645
GATC	125.6	100.0	100.0	617210
GATM	140.6	100.0	100.0	602360
GBF1	130.6	98.3	97.7	No OMIM phenotype
GDAP1	141.3	99.7	98.4	606598
GFER	89.3	99.8	97.6	600924

GFM1	110.9	99.7	98.7	606639
GFM2	124.8	98.1	93.7	606544
GLRX5	118.9	97.2	89.6	609588
GLUD1	64.6	96.4	84.4	138130
GMPR	118.2	100.0	99.9	139265
GOT2	74.6	94.6	87.0	138150
GPT2	123.8	99.4	95.3	138210
GTPBP2	128.9	99.8	98.5	607434
GTPBP3	176.4	100.0	99.9	608536
HACE1	146.1	99.7	99.3	610876
HADHA	70.8	95.5	88.3	600890
HADHB	73.3	97.7	87.0	143450
HARS2	126.8	100.0	99.5	600783
HCCS	99.1	99.3	96.1	300056
HIBCH	70.8	98.2	84.5	610690
HLCS	153.7	100.0	100.0	609018
HPDL	208.5	100.0	100.0	619026;619027
HSD17B10	103.8	99.9	98.3	300256
HSPA9	76.1	87.1	82.8	600548
HSPD1	81.1	96.7	90.0	118190
HTRA2	120.2	100.0	99.6	606441
IARS2	144.8	99.9	99.8	612801
IBA57	129.2	95.4	91.7	616451;615330
ISCA1	44.9	89.5	76.2	611006
ISCA2	110.4	99.8	96.5	615317
ISCU	115.9	100.0	100.0	611911
KARS1	109.3	99.9	98.9	601421
KIF1A	116.1	97.4	95.3	601255
LACTB	108.6	99.4	95.1	608440
LARS2	123.2	100.0	100.0	604544

LIAS	130.5	99.8	98.9	607031
LIPT1	156.1	99.7	99.5	610284
LIPT2	68.5	98.4	82.4	617659
LONP1	146.9	100.0	99.9	600373
LRPPRC	130.9	99.7	99.3	607544
LYRM4	90.7	66.7	65.6	613311
LYRM7	63.5	95.6	86.5	615831
MARS2	159.8	100.0	100.0	609728
MCUR1	57.9	99.1	91.3	616952
MDH1	100.1	99.7	99.1	154200
MDH2	102.0	98.0	98.0	154100
MECR	104.1	100.0	98.7	608205
MFF	88.3	93.9	89.4	614785
MFN2	118.1	100.0	99.8	608507
MGME1	150.6	100.0	99.9	615084
MICOS13	80.3	100.0	98.9	616658
MICU1	112.0	97.3	92.2	605084
MICU2	54.9	96.7	92.5	610632
MIEF2	122.5	100.0	99.1	615498
MIPEP	97.2	99.5	97.1	602241
MPC1	137.1	100.0	99.6	614741
MPV17	93.4	100.0	98.7	137960
MRM2	114.2	100.0	98.9	606906
MRPL12	131.7	100.0	99.1	602375
MRPL24	141.3	100.0	100.0	No OMIM phenotype
MRPL3	62.7	91.7	82.1	607118
MRPL40	97.9	99.5	91.6	605089
MRPL44	110.3	99.5	97.4	611849
MRPL57	178.9	100.0	100.0	611997
MRPS14	203.4	100.0	100.0	611978

MRPS16	130.6	100.0	98.8	609204
MRPS2	157.5	99.6	97.0	611971
MRPS22	127.4	99.7	98.3	605810
MRPS23	133.7	99.7	98.8	611985
MRPS25	133.1	100.0	99.8	611987
MRPS28	132.3	87.6	86.6	611990
MRPS34	121.1	98.6	93.3	611994
MRPS36	60.4	94.0	75.2	611996
MRPS7	136.9	100.0	100.0	611974
MRRF	129.7	100.0	100.0	604602
MSTO1	103.9	99.0	96.3	617619
MTFMT	138.9	99.9	99.5	611766
MTO1	138.3	90.9	88.8	614702
MTPAP	131.9	99.1	94.1	613669
MTX2	108.1	98.2	88.5	No OMIM phenotype
NARS2	116.3	97.9	97.1	612803
NAXD	131.1	100.0	99.9	615910
NAXE	80.6	100.0	98.6	608862
NDUFA1	185.7	99.8	99.3	300078
NDUFA10	131.5	99.9	98.6	603835
NDUFA11	118.1	100.0	99.8	612638
NDUFA12	173.0	99.6	99.6	614530
NDUFA13	112.3	92.2	90.0	609435
NDUFA2	140.6	100.0	100.0	602137
NDUFA3	139.4	88.7	88.0	603832
NDUFA4	83.7	99.1	96.5	603833
NDUFA5	66.0	92.3	75.3	601677
NDUFA6	198.5	100.0	100.0	602138
NDUFA7	103.9	100.0	99.7	602139
NDUFA8	135.2	100.0	97.3	603359



NDUFA9	102.5	99.3	95.2	603834
NDUFAB1	114.1	98.9	91.9	603836
NDUFAB1	116.6	100.0	100.0	606934
NDUFAB2	49.9	91.0	77.5	609653
NDUFAB3	128.1	100.0	99.9	612911
NDUFAB4	104.6	99.6	96.9	611776
NDUFAB5	119.6	99.7	99.1	612360
NDUFAB6	95.0	99.3	96.9	612392
NDUFAB7	101.5	99.8	99.3	252010
NDUFAB8	38.3	62.6	61.7	618461
NDUFB1	52.4	67.8	54.3	603837
NDUFB10	161.5	100.0	100.0	603843
NDUFB11	100.6	99.1	94.8	300403
NDUFB2	87.4	100.0	99.4	603838
NDUFB3	30.8	88.6	71.0	603839
NDUFB4	101.6	87.2	84.9	603840
NDUFB5	98.9	100.0	100.0	603841
NDUFB6	47.3	97.2	84.7	603322
NDUFB7	86.5	99.9	97.4	603842
NDUFB8	103.7	100.0	99.5	602140
NDUFB9	99.8	96.1	91.5	601445
NDUFC1	87.2	99.5	99.3	603844
NDUFC2	58.9	99.1	91.9	603845
NDUFS1	161.0	99.9	99.1	157655
NDUFS2	98.4	100.0	100.0	602985
NDUFS3	127.5	90.7	90.6	603846
NDUFS4	156.0	99.7	99.7	602694
NDUFS5	120.6	100.0	100.0	603847
NDUFS6	115.6	100.0	99.8	603848
NDUFS7	146.6	100.0	99.7	601825

NDUFS8	140.2	100.0	99.1	602141
NDUFV1	138.5	99.0	97.0	161015
NDUFV2	69.8	85.8	78.7	600532
NDUFV3	124.0	99.9	98.4	602184
NFS1	73.2	89.2	83.6	603485
NFU1	62.6	98.7	87.7	608100
NGLY1	142.3	99.8	99.7	610661
NME3	138.9	96.1	91.0	601817
NR2F1	188.1	100.0	100.0	132890
NSUN3	183.4	100.0	100.0	619012
NUBPL	94.2	99.5	96.9	613621
OGDH	165.6	100.0	99.8	613022
OPA1	128.7	99.5	96.7	605290
OPA3	134.8	100.0	99.5	606580
OTX2	124.0	100.0	99.0	600037
OXA1L	133.8	100.0	99.4	601066
PANK2	159.3	100.0	99.7	606157
PARS2	174.5	100.0	100.0	612036
PC	158.4	99.7	98.0	608786
PDE2A	114.0	100.0	99.5	No OMIM phenotype
PDHA1	85.2	98.8	95.9	300502
PDHB	111.0	99.2	96.8	179060
PDHX	143.5	99.8	99.6	608769
PDK1	124.7	99.7	99.2	605213
PDK2	148.8	100.0	100.0	602525
PDK3	101.8	98.8	95.5	602526
PDK4	116.9	99.7	99.5	602527
PDP1	129.4	100.0	100.0	605993
PDSS1	110.5	95.2	87.8	607429
PDSS2	112.7	98.4	94.3	610564

PET100	88.7	100.0	99.2	220110
PET117	101.2	100.0	100.0	614771
PIGA	73.9	91.6	82.5	300868
PISD	161.8	100.0	99.7	612770
PITRM1	104.7	98.2	96.2	619405
PLA2G6	111.6	92.1	90.7	610217;256600;612953
PLPBP	90.1	95.1	88.9	604436
PMPCA	105.8	97.6	93.5	213200
PMPCB	127.4	99.9	99.2	603131
PNPLA8	118.9	99.7	99.5	612123
PNPT1	54.0	96.9	86.1	610316
POLG	113.2	99.9	98.8	174763
POLG2	213.3	99.3	97.2	604983
POLR2A	162.8	100.0	100.0	180660
POLRMT	52.0	85.5	65.9	601778
PPA2	87.0	97.3	88.6	609988
PPCS	130.5	100.0	99.1	609853
PRKAA1	142.9	99.8	99.6	602739
PRPS1	103.4	86.4	86.3	300661
PTCD3	87.2	99.2	96.9	614918
PTRH2	223.0	100.0	100.0	616263
PUS1	117.5	99.9	98.0	608109
PYCR1	95.6	100.0	98.2	179035
PYCR2	127.2	100.0	99.3	616420
PYROXD1	49.7	92.1	78.7	617258
QRSL1	90.0	98.6	92.8	617209
RARS2	104.3	99.7	98.6	611524
RMND1	156.8	99.7	97.2	614917
RNASEH1	103.1	98.7	95.8	604123
RRM1	124.0	99.9	99.5	180410

RRM2B	147.5	100.0	99.8	604712
RTN4IP1	78.6	99.6	97.3	616732
RYR1	121.5	97.1	94.0	180901
SACS	151.3	99.9	99.9	604490
SAMHD1	137.6	98.5	97.9	606754
SARS2	115.0	95.7	94.5	612804
SCO1	93.4	97.6	94.4	603644
SCO2	117.1	100.0	100.0	604272
SCP2	111.6	99.9	97.9	184755
SDHA	94.3	84.5	77.9	600857
SDHAF1	49.2	100.0	98.4	612848
SDHB	123.9	100.0	100.0	185470
SDHD	38.8	53.8	49.0	602690
SERAC1	108.9	99.6	99.5	612073
SFXN4	102.7	99.6	97.4	615578
SLC19A2	95.1	100.0	98.5	603941
SLC19A3	130.4	97.8	97.0	606152
SLC25A1	91.4	96.9	89.8	190315
SLC25A10	70.0	76.4	70.3	606794
SLC25A12	147.0	100.0	99.2	603667
SLC25A13	110.8	100.0	99.4	603859
SLC25A19	84.3	99.9	98.0	606521
SLC25A21	121.8	100.0	99.6	607571
SLC25A22	117.3	99.2	96.5	609302
SLC25A24	128.8	99.3	98.8	608744
SLC25A3	126.4	99.7	96.9	600370
SLC25A32	123.9	100.0	100.0	610815
SLC25A38	98.1	97.4	93.3	610819
SLC25A4	126.5	100.0	99.8	103220
SLC25A42	133.3	97.1	94.3	610823

SLC25A46	184.6	99.7	98.6	616505
SLC39A8	126.5	100.0	99.7	608732
SLC52A2	171.9	100.0	100.0	607882
SLC52A3	127.1	100.0	100.0	613350
SOD2	195.5	100.0	100.0	147460
SPART	137.8	99.7	96.4	607111
SPATA5	137.1	99.8	99.5	613940
SPG7	104.5	90.4	86.7	602783
SQOR	89.5	100.0	98.0	617658
SQSTM1	120.9	99.8	97.8	601530
SSBP1	56.3	99.1	94.1	600439
STAC3	118.7	100.0	100.0	615521
STAT2	106.1	100.0	99.4	600556
STXBP1	105.8	96.8	96.2	602926
SUCLA2	55.1	88.8	79.4	603921
SUCLG1	102.4	100.0	99.7	611224
SUCLG2	55.7	91.7	79.1	603922
SURF1	81.5	89.5	88.1	185620
SZT2	144.1	99.6	99.3	615463
TACO1	86.6	98.9	93.7	612958
TANGO2	127.0	100.0	99.3	616830
TAOK1	126.8	99.6	97.5	610266
TARS2	94.6	99.9	98.8	612805
TAZ	104.7	99.3	93.7	300394
TDP2	166.5	99.6	99.5	605764
TFAM	75.5	98.0	78.5	617156
TFB2M	81.2	100.0	98.5	607055
THG1L	122.6	100.0	100.0	618800
TIMM22	82.3	100.0	99.2	607251
TIMM44	165.5	100.0	99.9	605058

TIMM50	126.6	98.4	95.0	607381
TIMM8A	50.9	96.2	83.1	300356
TIMMDC1	150.2	99.9	99.8	615534
TK2	104.4	99.0	96.0	188250
TMEM126A	105.6	95.4	80.0	612988
TMEM126B	84.3	99.6	97.2	252010
TMEM186	132.0	100.0	100.0	-
TMEM65	59.7	89.3	83.2	616609
TMEM70	116.7	98.4	94.6	612418
TMX2	130.2	100.0	99.2	616715
TOMM70	111.7	99.9	99.3	606081
TOP3A	119.9	99.6	96.5	601243
TPK1	99.5	99.5	97.2	606370
TRAPPC2L	186.5	100.0	100.0	610970
TRIT1	113.2	100.0	100.0	617873
TRMT10C	124.9	100.0	99.9	615423
TRMT5	186.4	99.8	99.1	611023
TRMU	100.4	99.9	99.6	610230
TRNT1	84.7	99.7	97.4	612907
TSFM	121.7	100.0	99.3	604723
TTC19	73.2	83.8	74.1	613814
TUFM	154.7	99.9	97.6	602389
TWNK	173.4	100.0	99.9	606486
TXN2	57.0	100.0	98.5	616811
TYMP	111.2	100.0	99.4	131222
UQCC1	94.0	100.0	100.0	611797
UQCC2	106.7	99.9	98.5	614461
UQCC3	103.7	100.0	97.5	616111
UQCR10	167.8	100.0	100.0	610843
UQCR11	195.0	100.0	100.0	609711

UQCRB	107.2	97.7	92.1	191330
UQCRC1	129.5	99.8	98.3	191328
UQCRC2	113.8	100.0	98.8	191329
UQCRFS1	106.1	94.1	88.8	191327
UQCRH	114.0	100.0	97.3	613844
UQCRQ	113.8	100.0	100.0	612080
VARS2	122.4	100.0	99.0	612802
VPS13D	142.0	100.0	99.4	608877
WARS2	136.3	100.0	99.8	604733
WDR45	71.6	98.2	92.0	300526
YARS2	171.1	99.9	99.4	610957
YME1L1	109.3	98.9	93.7	607472

*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.*