

WES MITOCHONDRIAL DISORDERS DG 3.1

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AARS2	131.5	100.0	99.4	612035
ABAT	105.5	100.0	99.4	613163
ACAD9	141.7	100.0	99.9	611103
ACO2	122.9	96.3	90.3	100850
ACTA1	86.4	99.6	92.3	102610
ADAMTS10	125.3	99.9	98.5	608990
ADCK2	168.4	100.0	100.0	-
ADPRS	159.0	100.0	99.8	610624
AFG3L2	115.3	95.0	91.1	604581
AGK	123.5	90.6	88.6	610345
AIFM1	117.3	99.9	98.8	300169
ALDH1B1	203.5	100.0	100.0	100670
ALKBH1	108.3	100.0	99.9	605345
ANO10	131.5	99.8	97.9	613726
APOO	69.8	85.7	75.3	No OMIM phenotype
APTX	122.7	94.9	92.4	606350
ARL2	133.7	100.0	100.0	601175
ARNT2	147.3	100.0	100.0	606036
ATAD1	77.5	99.6	95.1	614452
ATAD3A	84.8	91.9	83.2	612316
ATAD3B	85.2	92.1	79.2	612317
ATP13A2	138.1	100.0	99.5	610513
ATP5F1A	84.3	95.2	87.6	164360
ATP5F1B	124.6	100.0	97.8	102910
ATP5F1C	87.6	98.0	92.2	108729

ATP5F1D	72.7	96.2	89.3	603150
ATP5F1E	175.0	100.0	100.0	606153
ATP5IF1	193.9	100.0	100.0	614981
ATP5MC1	99.2	100.0	99.9	603192
ATP5MC2	79.1	100.0	98.1	603193
ATP5MC3	136.5	100.0	100.0	602736
ATP5MD	19.8	82.9	42.8	615204
ATP5ME	83.6	100.0	100.0	601519
ATP5MF	150.9	100.0	99.4	-
ATP5MG	137.0	100.0	100.0	617473
ATP5MGL	197.1	100.0	100.0	613213
ATP5PB	68.8	98.9	90.4	603270
ATP5PD	98.0	97.3	80.9	618121
ATP5PF	69.4	100.0	97.5	603152
ATP5PO	114.9	99.9	98.0	600828
ATPAF1	81.6	78.3	70.0	608917
ATPAF2	118.7	100.0	100.0	608918
BCAP31	80.0	92.6	83.2	300475
BCS1L	158.0	100.0	100.0	603647
BOLA1	91.4	100.0	100.0	613181
BOLA2	115.5	100.0	100.0	613182
BOLA3	50.8	99.4	90.2	613183
C12orf65	122.8	99.8	98.5	613541
C19orf12	122.6	100.0	99.8	614297
C1QBP	76.2	86.9	77.3	601269
CA5A	106.3	87.4	85.2	114761
CARS2	131.3	100.0	100.0	612800
CEP89	159.0	96.0	94.5	615470
CFAP58	109.9	99.9	98.7	No OMIM phenotype
CHCHD10	25.2	59.1	43.9	615903

CHCHD2	76.6	98.4	83.8	616710
CHKB	114.9	100.0	99.7	612395
CISD2	135.4	83.4	83.4	611507
CLPB	135.7	94.9	94.9	616271
CLPP	147.3	100.0	99.1	601119
COA1	116.1	100.0	100.0	614769
COA3	156.1	100.0	100.0	614775
COA5	68.9	99.1	88.9	613920
COA6	106.3	99.9	98.4	614772
COA7	142.6	100.0	100.0	615623
COA8	92.5	81.9	80.7	616003
COASY	181.4	100.0	100.0	609855
COQ2	113.1	98.0	95.3	609825
COQ4	98.8	90.9	89.3	612898
COQ5	196.7	100.0	100.0	616359
COQ6	143.6	99.9	98.4	614647
COQ7	161.7	100.0	99.8	601683
COQ8A	164.9	100.0	99.5	606980
COQ8B	102.1	100.0	99.3	615567
COQ9	79.4	100.0	97.9	612837
COX10	211.4	100.0	100.0	602125
COX14	115.3	100.0	100.0	614478
COX15	106.0	99.9	98.8	603646
COX16	85.6	99.1	96.8	618064
COX20	72.8	97.8	88.3	614698
COX411	121.7	100.0	100.0	123864
COX412	127.4	100.0	100.0	607976
COX5A	31.5	74.7	47.1	603773
COX5B	154.2	100.0	100.0	123866
COX6A1	177.4	100.0	99.5	602072

COX6A2	51.6	99.2	93.7	602009
COX6B1	189.5	100.0	100.0	124089
COX6B2	81.1	100.0	99.8	220110
COX6C	110.4	100.0	97.4	124090
COX7A1	145.2	100.0	99.9	123995
COX7A2	117.2	100.0	99.8	123996
COX7B	52.2	78.2	49.4	603792
COX7B2	150.1	100.0	100.0	609811
COX7C	39.4	99.3	86.9	603774
COX8A	111.3	100.0	100.0	123870
COX8C	152.7	100.0	99.9	220110
CP	127.1	94.8	88.9	117700
CRAT	126.1	100.0	99.8	600184
CTBP1	96.7	93.2	86.9	602618
CYC1	163.2	97.5	89.2	123980
CYCS	78.6	99.1	94.9	123970
DARS2	143.6	94.9	94.3	610956
DCAF17	107.3	98.9	93.3	612515
DDHD1	164.0	97.9	95.8	614603
DES	119.2	100.0	99.7	125660
DGUOK	124.5	100.0	99.4	601465
DHTKD1	145.8	99.9	98.9	614984
DLAT	111.4	100.0	99.7	608770
DLD	135.6	100.0	99.7	238331
DLST	97.1	96.7	90.3	126063
DMAC1	64.1	100.0	100.0	617261
DMAC2	143.6	98.3	98.3	617262
DMAC2L	117.5	100.0	100.0	603152
DNA2	155.0	99.8	98.3	601810
DNAJA3	142.1	98.6	96.0	608382

DNAJC19	139.7	98.9	96.2	608977
DNAJC3	148.3	100.0	99.7	601184
DNAJC30	173.0	100.0	100.0	618202
DNM1L	139.3	99.9	98.5	603850
EARS2	105.9	99.8	97.7	612799
ECHS1	108.6	99.9	99.0	602292
ECSIT	142.0	100.0	99.9	608388
EHHADH	164.6	100.0	100.0	607037
ELAC2	124.7	100.0	99.7	605367
ERAL1	173.9	100.0	99.7	607435
ETFDH	141.8	100.0	99.8	231675
ETHE1	96.2	99.9	97.4	608451
FA2H	96.4	92.0	83.1	611026
FARS2	206.7	100.0	100.0	614946
FARSB	94.2	98.8	94.6	609690
FASTKD2	141.3	99.8	98.9	612322
FBXL4	200.7	100.0	100.0	605654
FDX2	137.1	100.0	100.0	614585
FDXR	126.5	100.0	99.3	103270
FH	147.0	92.1	88.3	606945
FOXRED1	136.0	100.0	99.9	613622
FTL	144.1	98.5	89.4	134790
FXN	72.3	95.5	80.1	606829
GARS1	154.4	99.9	99.1	600287
GATB	128.0	100.0	99.7	603645
GATC	137.1	100.0	100.0	617210
GATM	156.2	100.0	100.0	602360
GBF1	142.1	98.3	98.0	No OMIM phenotype
GDAP1	185.1	99.8	99.3	606598
GFER	91.5	99.6	93.9	600924

GFM1	131.2	99.9	99.4	606639
GFM2	137.5	98.9	95.2	606544
GLRX5	129.2	97.3	89.1	609588
GLUD1	72.8	94.2	82.9	138130
GMPR	135.9	100.0	100.0	139265
GOT2	95.7	97.5	90.9	138150
GPT2	136.1	99.2	93.6	138210
GTPBP2	140.5	100.0	99.3	607434
GTPBP3	163.2	100.0	99.8	608536
HACE1	171.9	100.0	99.3	610876
HADHA	87.5	97.2	91.6	600890
HADHB	90.7	98.8	89.7	143450
HARS2	148.6	100.0	100.0	600783
HCCS	110.5	99.8	97.6	300056
HIBCH	83.8	98.2	88.5	610690
HLCS	179.3	100.0	100.0	609018
HPDL	200.6	100.0	100.0	619026;619027
HSD17B10	109.8	100.0	99.1	300256
HSPA9	97.6	88.5	84.5	600548
HSPD1	91.9	98.8	93.7	118190
HTRA2	138.0	100.0	99.9	606441
IARS2	170.4	100.0	99.9	612801
IBA57	117.3	93.7	90.1	616451;615330
ISCA1	54.5	94.2	85.9	611006
ISCA2	126.3	100.0	98.8	615317
ISCU	135.3	100.0	100.0	611911
KARS1	133.2	100.0	99.9	601421
KIF1A	114.7	97.4	95.2	601255
LACTB	128.2	99.5	92.6	608440
LARS2	143.2	100.0	100.0	604544

LIAS	151.6	100.0	99.1	607031
LIPT1	227.5	100.0	99.9	610284
LIPT2	70.0	94.9	75.2	617659
LONP1	141.9	100.0	99.8	600373
LRPPRC	151.5	99.9	99.1	607544
LYRM4	107.2	68.5	66.2	613311
LYRM7	76.8	95.9	86.2	615831
MARS2	170.6	100.0	100.0	609728
MCUR1	68.3	99.5	91.5	616952
MDH1	112.8	100.0	99.0	154200
MDH2	116.9	98.0	97.9	154100
MECR	115.4	100.0	98.9	608205
MFF	101.3	94.3	89.9	614785
MFN2	138.2	100.0	99.9	608507
MGME1	183.0	100.0	100.0	615084
MICOS13	81.0	100.0	99.7	616658
MICU1	126.1	98.9	95.2	605084
MICU2	66.4	97.2	91.8	610632
MIEF2	123.1	100.0	99.0	615498
MIPEP	118.5	99.2	96.5	602241
MPC1	164.8	100.0	99.6	614741
MPV17	97.4	100.0	97.2	137960
MRM2	134.9	100.0	99.4	606906
MRPL12	130.9	100.0	98.2	602375
MRPL24	152.7	100.0	99.9	No OMIM phenotype
MRPL3	76.1	93.2	87.2	607118
MRPL40	94.0	99.9	96.1	605089
MRPL44	140.0	99.9	98.7	611849
MRPL57	168.3	100.0	99.8	611997
MRPS14	208.4	100.0	100.0	611978

MRPS16	151.3	100.0	99.6	609204
MRPS2	167.5	99.6	96.9	611971
MRPS22	153.4	99.8	99.1	605810
MRPS23	158.3	100.0	99.6	611985
MRPS25	146.9	100.0	99.9	611987
MRPS28	158.7	89.9	86.8	611990
MRPS34	133.8	97.6	92.0	611994
MRPS36	69.7	95.2	77.6	611996
MRPS7	167.3	100.0	100.0	611974
MRRF	161.6	100.0	100.0	604602
MSTO1	119.3	99.6	96.8	617619
MTFMT	165.4	100.0	99.8	611766
MTO1	165.3	91.3	90.4	614702
MTPAP	151.0	99.5	96.1	613669
MTX2	133.6	98.1	91.5	No OMIM phenotype
NARS2	145.9	98.3	97.4	612803
NAXD	142.8	100.0	100.0	615910
NAXE	85.1	100.0	99.8	608862
NDUFA1	196.4	99.9	99.3	300078
NDUFA10	145.3	99.8	98.6	603835
NDUFA11	117.9	100.0	100.0	612638
NDUFA12	198.9	100.0	100.0	614530
NDUFA13	122.5	92.2	89.2	609435
NDUFA2	127.1	100.0	100.0	602137
NDUFA3	139.8	89.1	87.9	603832
NDUFA4	93.0	100.0	96.4	603833
NDUFA5	79.8	96.5	81.8	601677
NDUFA6	206.0	100.0	100.0	602138
NDUFA7	119.6	100.0	99.9	602139
NDUFA8	163.5	100.0	99.0	603359

NDUFA9	115.5	99.9	96.5	603834
NDUFAB1	134.2	99.6	94.9	603836
NDUFAF1	135.8	100.0	100.0	606934
NDUFAF2	63.6	95.0	83.4	609653
NDUFAF3	129.2	100.0	99.9	612911
NDUFAF4	141.6	99.8	98.2	611776
NDUFAF5	139.0	100.0	99.5	612360
NDUFAF6	106.2	100.0	96.8	612392
NDUFAF7	119.7	100.0	99.8	252010
NDUFAF8	38.0	62.6	61.1	618461
NDUFB1	59.9	78.2	57.8	603837
NDUFB10	170.8	100.0	100.0	603843
NDUFB11	108.7	99.5	96.5	300403
NDUFB2	99.4	100.0	100.0	603838
NDUFB3	34.6	95.8	80.5	603839
NDUFB4	129.5	90.2	85.2	603840
NDUFB5	122.0	100.0	100.0	603841
NDUFB6	58.3	98.4	88.4	603322
NDUFB7	87.1	99.8	94.8	603842
NDUFB8	114.9	100.0	99.9	602140
NDUFB9	106.8	96.5	92.5	601445
NDUFC1	91.7	99.6	99.4	603844
NDUFC2	62.1	99.8	96.0	603845
NDUFS1	178.7	100.0	99.5	157655
NDUFS2	118.5	100.0	100.0	602985
NDUFS3	141.3	90.7	90.6	603846
NDUFS4	190.8	100.0	99.4	602694
NDUFS5	141.8	100.0	100.0	603847
NDUFS6	138.2	100.0	99.9	603848
NDUFS7	125.4	100.0	99.2	601825

NDUFS8	134.0	100.0	99.4	602141
NDUFV1	158.4	98.0	96.1	161015
NDUFV2	84.9	86.9	76.9	600532
NDUFV3	153.2	99.8	98.0	602184
NFS1	84.2	87.9	84.0	603485
NFU1	72.1	98.8	90.8	608100
NGLY1	166.1	100.0	99.8	610661
NME3	128.0	93.1	88.5	601817
NR2F1	182.5	100.0	100.0	132890
NSUN3	215.1	100.0	100.0	8
NUBPL	111.9	99.7	98.4	613621
OGDH	185.9	100.0	99.9	613022
OPA1	155.2	99.6	97.6	605290
OPA3	124.4	100.0	99.0	606580
OTX2	152.3	100.0	99.7	600037
OXA1L	167.6	100.0	99.8	601066
PANK2	185.6	100.0	99.3	606157
PARS2	188.8	100.0	100.0	612036
PC	161.8	99.8	97.3	608786
PDE2A	114.8	100.0	99.9	No OMIM phenotype
PDHA1	102.0	99.4	97.1	300502
PDHB	132.1	99.1	97.5	179060
PDHX	159.4	99.9	99.4	608769
PDK1	143.5	99.9	98.5	605213
PDK2	156.5	100.0	100.0	602525
PDK3	122.5	99.5	97.2	602526
PDK4	133.5	100.0	99.3	602527
PDP1	151.0	100.0	100.0	605993
PDSS1	129.8	94.7	87.6	607429
PDSS2	125.9	99.8	97.1	610564

PET100	107.1	100.0	99.6	220110
PET117	117.9	100.0	100.0	614771
PIGA	85.7	93.8	86.7	300868
PISD	168.8	100.0	100.0	612770
PITRM1	122.8	98.4	96.1	6
PLA2G6	112.9	92.2	90.7	610217;256600;612953
PLPBP	111.9	98.2	90.1	604436
PMPCA	123.2	97.7	94.2	213200
PMPCB	145.8	100.0	99.7	603131
PNPLA8	140.1	100.0	99.8	612123
PNPT1	63.9	97.7	89.7	610316
POLG	111.5	100.0	99.3	174763
POLG2	235.4	99.6	98.0	604983
POLR2A	182.8	100.0	100.0	180660
POLRMT	48.6	81.9	63.4	601778
PPA2	94.4	98.7	94.0	609988
PPCS	139.9	99.8	99.5	609853
PRKAA1	176.8	100.0	99.5	602739
PRPS1	118.4	86.4	86.4	300661
PTCD3	100.2	99.2	97.6	614918
PTRH2	259.0	100.0	100.0	616263
PUS1	118.0	100.0	99.5	608109
PYCR1	94.2	99.9	97.7	179035
PYCR2	118.2	100.0	99.1	616420
PYROXD1	56.6	95.2	83.9	617258
QRSL1	101.2	99.2	93.9	617209
RARS2	122.7	100.0	99.8	611524
RMND1	165.2	100.0	98.6	614917
RNASEH1	109.0	98.5	95.3	604123
RRM1	142.7	100.0	99.5	180410

RRM2B	163.8	100.0	99.7	604712
RTN4IP1	96.9	99.9	98.7	616732
RYR1	126.2	96.9	93.9	180901
SACS	174.6	100.0	99.9	604490
SAMHD1	162.2	98.7	98.4	606754
SARS2	118.3	95.8	94.6	612804
SCO1	115.4	97.1	93.8	603644
SCO2	107.3	100.0	100.0	604272
SCP2	130.8	100.0	99.2	184755
SDHA	100.0	85.8	80.4	600857
SDHAF1	40.4	99.9	93.2	612848
SDHB	146.3	100.0	100.0	185470
SDHD	48.5	54.0	51.6	602690
SERAC1	134.6	99.9	99.5	612073
SFXN4	128.8	99.9	98.9	615578
SLC19A2	110.3	100.0	99.7	603941
SLC19A3	156.5	97.8	97.6	606152
SLC25A1	97.8	95.8	88.6	190315
SLC25A10	68.1	76.2	69.3	606794
SLC25A12	187.5	99.9	99.5	603667
SLC25A13	146.4	100.0	99.7	603859
SLC25A19	94.0	100.0	98.5	606521
SLC25A21	142.7	100.0	99.7	607571
SLC25A22	115.0	98.6	95.8	609302
SLC25A24	146.3	99.4	99.3	608744
SLC25A3	138.4	99.8	98.0	600370
SLC25A32	145.3	100.0	100.0	610815
SLC25A38	112.2	97.9	95.3	610819
SLC25A4	144.8	100.0	100.0	103220
SLC25A42	131.7	96.5	93.2	610823

SLC25A46	199.3	99.7	97.3	616505
SLC39A8	145.0	100.0	99.7	608732
SLC52A2	170.8	100.0	100.0	607882
SLC52A3	125.5	100.0	100.0	613350
SOD2	228.2	100.0	100.0	147460
SPART	153.2	99.7	96.8	607111
SPATA5	167.7	100.0	99.7	613940
SPG7	116.2	88.2	86.2	602783
SQOR	108.8	100.0	97.8	617658
SQSTM1	125.7	98.8	95.5	601530
SSBP1	68.3	99.8	97.6	600439
STAC3	132.6	100.0	100.0	615521
STAT2	115.3	100.0	99.9	600556
STXBP1	120.1	96.8	96.5	602926
SUCLA2	62.7	89.5	82.2	603921
SUCLG1	122.6	99.9	99.8	611224
SUCLG2	72.5	96.7	86.3	603922
SURF1	94.9	89.4	88.2	185620
SZT2	153.3	99.6	99.5	615463
TACO1	96.1	98.4	93.0	612958
TANGO2	133.1	100.0	99.3	616830
TAOK1	152.4	99.5	97.9	610266
TARS2	107.1	100.0	99.3	612805
TAZ	101.2	99.1	95.5	300394
TDP2	198.6	100.0	99.4	605764
TFAM	86.0	97.5	83.5	617156
TFB2M	95.6	100.0	99.1	607055
THG1L	160.5	100.0	100.0	7
TIMM22	91.7	100.0	99.7	607251
TIMM44	166.9	100.0	99.4	605058

TIMM50	129.7	98.3	94.4	607381
TIMM8A	62.7	98.1	90.6	300356
TIMMDC1	189.0	100.0	100.0	615534
TK2	119.9	99.2	96.3	188250
TMEM126A	125.2	96.3	84.4	612988
TMEM126B	100.8	99.8	97.4	252010
TMEM186	143.2	100.0	100.0	-
TMEM65	64.8	88.0	81.3	616609
TMEM70	130.2	98.0	93.9	612418
TMX2	150.7	100.0	99.8	616715
TOMM70	128.7	100.0	99.8	606081
TOP3A	132.1	100.0	98.7	601243
TPK1	121.5	99.8	99.0	606370
TRAPPC2L	213.9	100.0	100.0	610970
TRIT1	133.0	100.0	100.0	617873
TRMT10C	157.4	100.0	100.0	615423
TRMT5	217.7	100.0	99.3	611023
TRMU	103.5	100.0	100.0	610230
TRNT1	108.4	99.5	96.5	612907
TSFM	139.8	100.0	99.5	604723
TTC19	89.7	81.5	73.8	613814
TUFM	158.0	100.0	99.0	602389
TWNK	202.8	100.0	100.0	606486
TXN2	74.6	100.0	100.0	616811
TYMP	97.1	100.0	97.0	131222
UQCC1	115.9	100.0	99.9	611797
UQCC2	114.9	100.0	99.7	614461
UQCC3	123.3	100.0	98.7	616111
UQCR10	170.4	100.0	100.0	610843
UQCR11	217.1	100.0	100.0	609711

UQCRB	117.8	99.4	95.1	191330
UQCRC1	132.5	99.8	98.4	191328
UQCRC2	133.3	99.9	99.3	191329
UQCRFS1	121.6	91.9	84.9	191327
UQCRH	135.0	100.0	98.2	613844
UQCRQ	146.8	100.0	100.0	612080
VAR2	139.2	100.0	99.4	612802
VPS13D	171.0	100.0	99.7	608877
WARS2	140.8	100.0	99.4	604733
WDR45	82.5	98.1	92.4	300526
YARS2	194.6	100.0	99.8	610957
YME1L1	133.4	99.0	95.2	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.

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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors