MITOCHONDRIAL DISORDERS GENE PANEL DG 3.2.0 (427 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
AARS2	100	99,4	100	100	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABAT	99,9	97,8	100	100	GABA-transaminase deficiency, 613163
ACAD9	100	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACO2	94,1	86,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACTA1	98,2	89,5	100	100	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ADAMTS10	100	99,9	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADCK2	100	99,7	100	100	No OMIM disease ID
ADPRS	100	99,9	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	94,6	86,3	100	100	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGK	90,4	87,9	91,2	91,1	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AIFM1	99,9	97,8	100	100	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
ALDH1B1	100	100	100	100	No OMIM disease ID
ALKBH1	100	99,7	100	100	No OMIM disease ID
ANO10	99,2	96,6	100	100	Spinocerebellar ataxia, autosomal recessive 10, 613728
APOO	81,1	71,1	100	100	No OMIM disease ID

APTX	94,1	90,6	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARL2	100	99,7	100	100	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARNT2	100	100	100	99,5	?Webb-Dattani syndrome, 615926
ATAD1	99,1	91,6	100	100	Hyperekplexia 4, 618011
ATAD3A	91,4	86,7	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	91,1	83,2	100	100	No OMIM disease ID
ATP13A2	100	99,6	100	100	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP5F1A	92,2	83	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1B	99,8	97,7	100	100	No OMIM disease ID
ATP5F1C	95,5	89,3	100	100	No OMIM disease ID
ATP5F1D	97,4	91,8	100	100	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100	100	100	100	No OMIM disease ID
ATP5MC1	100	99,6	100	100	No OMIM disease ID
ATP5MC2	99,3	92,4	100	100	No OMIM disease ID
ATP5MC3	100	100	100	100	No OMIM disease ID
ATP5ME	100	100	100	100	No OMIM disease ID
ATP5MF	99,6	94,8	100	100	No OMIM disease ID
ATP5MG	100	100	100	100	No OMIM disease ID
ATP5MGL	100	100	100	100	No OMIM disease ID
ATP5MD	83,8	35,7	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	97	83,3	100	100	No OMIM disease ID
ATP5PD	89,2	67,8	100	100	No OMIM disease ID
ATP5PF	99,9	92,4	100	100	No OMIM disease ID
ATP5PO	100	98,1	100	99,7	No OMIM disease ID
ATPAF1	83,7	71,4	100	100	No OMIM disease ID
ATPAF2	100	99,9	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273

BCAP31	92,1	79,1	100	99,6	Deafness, dystonia, and cerebral hypomyelination, 300475
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BOLA1	100	99,9	100	100	No OMIM disease ID
BOLA2	100	100	100	100	No OMIM disease ID
BOLA3	99	86,7	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QBP	84,3	70,6	100	100	Combined oxidative phosphorylation deficiency 33, 617713
CA5A	87,6	85,6	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CARS2	100	100	100	99,5	Combined oxidative phosphorylation deficiency 27, 616672
CEP89	95,8	94,5	100	100	No OMIM disease ID
CFAP58	99,7	97,1	100	100	Spermatogenic failure 49, 619144
CHCHD10	57,8	42	100	100	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	93,7	78,7	100	100	Parkinson disease 22, autosomal dominant, 616710
CHKB	100	99,6	100	100	Muscular dystrophy, congenital, megaconial type, 602541
CISD2	83,4	83,4	100	100	Wolfram syndrome 2, 604928
CLPB	94,9	94	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100	99,5	100	100	Perrault syndrome 3, 614129
COA1	100	100	100	100	No OMIM disease ID
COA3	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	94,4	83	85,2	85,2	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	99,6	96,3	100	100	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100	100	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	81,9	80,8	93,7	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100	100	100	100	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COQ2	97,6	96,7	97,2	97,2	Coenzyme Q10 deficiency, primary, 1, 607426

COQ4	91	89,7	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100	100	100	100	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	99,9	98,5	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,6	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100	99,6	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,2	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	98,7	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100	99,9	100	100	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	99,9	97,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	99,3	96,9	100	99,9	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX20	95,7	82,4	100	99,9	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100	99,9	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	66	36	100	100	?Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100	100	100	100	No OMIM disease ID
COX6A1	100	99,9	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,8	94	100	100	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100	99,9	100	100	No OMIM disease ID
COX6C	99,5	94,3	100	99,9	No OMIM disease ID
COX7A1	100	99,9	100	100	No OMIM disease ID
COX7A2	100	98,8	100	99,9	No OMIM disease ID
COX7B	69,1	35,9	100	100	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100	100	100	100	No OMIM disease ID
COX7C	98,4	78,9	100	100	No OMIM disease ID
COX8A	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100	99,9	100	100	No OMIM disease ID

CP	92,6	85,2	100	99,9	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CRAT	100	99,9	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CTBP1	94,3	86,9	99,4	98,4	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CYC1	98,3	89,5	100	99,4	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	99,4	96,9	100	100	Thrombocytopenia 4, 612004
DARS2	94,8	93,8	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCAF17	98,5	93,4	100	100	Woodhouse-Sakati syndrome, 241080
DDHD1	98,5	96,5	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DES	100	99,6	100	100	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGUOK	99,9	98,8	100	100	Portal hypertension, noncirrhotic, 1, 617068 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHTKD1	99,8	98,8	100	100	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DLAT	99,8	99,3	100	99,9	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	99,9	99,7	100	99,9	Dihydrolipoamide dehydrogenase deficiency, 246900
DLST	95,7	87,7	100	100	Paragangliomas 7, 618475
DMAC1	100	99,9	100	100	No OMIM disease ID
DMAC2	98,3	98,3	100	100	No OMIM disease ID
DMAC2L	99,8	99,8	100	99,9	No OMIM disease ID
DNA2	99,6	96,9	100	100	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156
DNAJA3	98,5	96,1	100	100	No OMIM disease ID
DNAJC19	99,3	92,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNAJC3	99,7	99,7	100	99,9	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100	100	100	100	Leber hereditary optic neuropathy, autosomal recessive, 619382
DNM1L	99,6	98,3	100	100	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
EARS2	99,8	98	100	100	Combined oxidative phosphorylation deficiency 12, 614924

ECHS1	100	99,4	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECSIT	100	100	100	100	No OMIM disease ID
EHHADH	100	100	100	100	?Fanconi renotubular syndrome 3, 615605
ELAC2	100	99,2	100	100	Combined oxidative phosphorylation deficiency 17, 615440
ERAL1	100	99,6	100	100	Perrault syndrome 6, 617565
ETFDH	99,8	99,4	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,3	93,3	100	100	Ethylmalonic encephalopathy, 602473
FA2H	92,4	82,6	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100	100	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSB	98	92,9	100	100	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	99,6	98,6	100	100	Combined oxidative phosphorylation deficiency 44, 618855
FBXL4	100	100	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FDX2	100	100	100	100	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100	98,6	100	100	Auditory neuropathy and optic atrophy, 617717
FH	93,2	87,2	100	100	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FOXRED1	100	99,6	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FTL	98,6	88,5	100	100	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	98,3	84,7	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GARS1	99,9	99,4	100	100	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GATB	100	99	100	100	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100	100	100	100	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBF1	98,3	97,7	100	100	No OMIM disease ID
GDAP1	99,7	98,4	100	100	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 Charcot-Marie-Tooth disease, recessive intermediate, A, 608340

					Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, type 4A, 214400
GFER	99,8	97,6	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	99,7	98,7	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,1	93,7	100	100	Combined oxidative phosphorylation deficiency 39, 618397
GLRX5	97,2	89,6	99,3	95,2	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLUD1	96,4	84,4	100	100	Hyperinsulinism-hyperammonemia syndrome, 606762
GMPR	100	99,9	100	100	No OMIM disease ID
GOT2	94,6	87	100	100	Developmental and epileptic encephalopathy 82, 618721
GPT2	99,4	95,3	100	100	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GTPBP2	99,8	98,5	100	100	Jaberi-Elahi syndrome, 617988
GTPBP3	100	99,9	100	100	Combined oxidative phosphorylation deficiency 23, 616198
HACE1	99,7	99,3	100	99,9	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADHA	95,5	88,3	100	100	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	97,7	87	100	99,9	Trifunctional protein deficiency, 609015
HARS2	100	99,5	100	99,8	Perrault syndrome 2, 614926
HCCS	99,3	96,1	100	100	Linear skin defects with multiple congenital anomalies 1, 309801
HIBCH	98,2	84,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HPDL	100	100	100	100	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HSD17B10	99,9	98,3	100	100	HSD10 mitochondrial disease, 300438
HSPA9	87,1	82,8	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	96,7	90	100	100	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100	99,6	100	100	3-methylglutaconic aciduria, type VIII, 617248
IARS2	99,9	99,8	100	100	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007

IBA57	95,4	91,7	100	100	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ISCA1	89,5	76,2	95,1	95,1	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	99,8	96,5	100	100	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100	100	100	100	Myopathy with lactic acidosis, hereditary, 255125
KARS1	99,9	98,9	100	100	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KIF1A	97,4	95,3	98	98	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
LACTB	99,4	95,1	100	99,9	No OMIM disease ID
LARS2	100	100	100	100	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIAS	99,8	98,9	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT1	99,7	99,5	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	98,4	82,4	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LONP1	100	99,9	100	100	CODAS syndrome, 600373
LRPPRC	99,7	99,3	100	99,9	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LYRM4	66,7	65,6	66,3	66,3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,6	86,5	100	99,2	Mitochondrial complex III deficiency, nuclear type 8, 615838
MARS2	100	100	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MCUR1	99,1	91,3	100	99,9	No OMIM disease ID
MDH1	99,7	99,1	100	100	?Developmental and epileptic encephalopathy 88, 618959
MDH2	98	98	100	100	Developmental and epileptic encephalopathy 51, 617339
MECR	100	98,7	100	100	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	93,9	89,4	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100	99,8	100	100	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Hereditary motor and sensory neuropathy VIA, 601152

MGME1	100	99,9	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MICOS13	100	98,9	100	99,9	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	97,3	92,2	100	100	Myopathy with extrapyramidal signs, 615673
MICU2	96,7	92,5	100	99,9	No OMIM disease ID
MIEF2	100	99,1	100	100	?Combined oxidative phosphorylation deficiency 49, 619024
MIPEP	99,5	97,1	100	100	Combined oxidative phosphorylation deficiency 31, 617228
MPC1	100	99,6	100	100	Mitochondrial pyruvate carrier deficiency, 614741
MPV17	100	98,7	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRM2	100	98,9	98,9	98,9	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100	99,1	100	100	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100	100	100	100	No OMIM disease ID
MRPL3	91,7	82,1	100	100	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	99,5	91,6	100	100	No OMIM disease ID
MRPL44	99,5	97,4	100	100	?Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	100	100	100	100	No OMIM disease ID
MRPS14	100	100	100	100	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100	98,8	100	100	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,6	97	100	100	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,7	98,3	100	100	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	99,7	98,8	100	100	?Combined oxidative phosphorylation defiency 46, 618952
MRPS25	100	99,8	82,7	82,7	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	87,6	86,6	86,6	86,6	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	98,6	93,3	100	100	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	94	75,2	100	100	No OMIM disease ID
MRPS7	100	100	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100	100	100	100	No OMIM disease ID
MSTO1	99	96,3	100	100	Myopathy, mitochondrial, and ataxia, 617675

MTFMT	99,9	99,5	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTO1	90,9	88,8	92,8	91,4	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	99,1	94,1	100	100	?Spastic ataxia 4, autosomal recessive, 613672
C12orf65	99	94,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTX2	98,2	88,5	100	100	Mandibuloacral dysplasia progeroid syndrome, 619127
NARS2	97,9	97,1	100	99,9	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAXD	100	99,9	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	98,6	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFA1	99,8	99,3	100	100	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	99,9	98,6	100	100	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100	99,8	100	99,9	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	99,6	99,6	100	100	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,2	90	100	100	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	88,7	88	92,2	88,4	No OMIM disease ID
NDUFA4	99,1	96,5	100	100	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	92,3	75,3	100	99,7	No OMIM disease ID
NDUFA6	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100	99,7	100	100	No OMIM disease ID
NDUFA8	100	97,3	100	100	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	99,3	95,2	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	98,9	91,9	100	100	No OMIM disease ID
NDUFAF1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	91	77,5	100	99,6	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,6	96,9	100	99,8	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	99,7	99,1	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238

NDUFAF6	99,3	96,9	100	99,9	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913
NDUFAF7	99,8	99,3	100	100	No OMIM disease ID
NDUFAF8	62,6	61,7	100	100	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	67,8	54,3	100	100	No OMIM disease ID
NDUFB10	100	100	100	100	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99,1	94,8	99,9	99,1	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100	99,4	100	100	No OMIM disease ID
NDUFB3	88,6	71	100	100	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	87,2	84,9	100	100	No OMIM disease ID
NDUFB5	100	100	100	100	No OMIM disease ID
NDUFB6	97,2	84,7	100	100	No OMIM disease ID
NDUFB7	99,9	97,4	100	100	No OMIM disease ID
NDUFB8	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	96,1	91,5	98,7	98,7	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	99,5	99,3	100	100	No OMIM disease ID
NDUFC2	99,1	91,9	100	100	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	99,9	99,1	100	99,9	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	92,8	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	99,7	99,7	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100	100	100	99,7	No OMIM disease ID
NDUFS6	100	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100	99,7	100	100	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100	99,1	100	100	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	99	97	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	85,8	78,7	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	99,9	98,4	100	100	No OMIM disease ID
NFS1	89,2	83,6	89,5	89,5	Combined oxidative phosphorylation deficiency 52, 619386

NFU1	98,7	87,7	100	100	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	99,8	99,7	100	100	Congenital disorder of deglycosylation, 615273
NME3	96,1	91	100	100	No OMIM disease ID
NR2F1	100	100	98,2	93	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NSUN3	100	100	100	100	Combined oxidative phosphorylation deficiency 48, 619012
NUBPL	99,5	96,9	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
OGDH	100	99,8	100	100	No OMIM disease ID
OPA1	99,5	96,7	100	99,9	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100	99,5	100	100	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OTX2	100	99	100	100	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
OXA1L	100	99,4	100	100	No OMIM disease ID
PANK2	100	99,7	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PARS2	100	100	100	100	Developmental and epileptic encephalopathy 75, 618437
PC	99,7	98	100	100	Pyruvate carboxylase deficiency, 266150
PDE2A	100	99,5	100	100	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDHA1	98,8	95,9	100	100	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99,2	96,8	100	100	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,8	99,6	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	99,7	99,2	100	99,9	No OMIM disease ID
PDK2	100	100	100	100	No OMIM disease ID
PDK3	98,8	95,5	100	99,8	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	99,7	99,5	100	99,9	No OMIM disease ID
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	95,2	87,8	97,4	97,4	Coenzyme Q10 deficiency, primary, 2, 614651

PDSS2	98,4	94,3	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PET100	100	99,2	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PIGA	91,6	82,5	100	99,8	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PISD	100	99,7	100	100	Liberfarb syndrome, 618889
PITRM1	98,2	96,2	100	100	Spinocerebellar ataxia, autosomal recessive 30, 619405
PLA2G6	92,1	90,7	92,3	92,3	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLPBP	95,1	88,9	100	99,9	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMPCA	97,6	93,5	100	100	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	99,9	99,2	100	100	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNPLA8	99,7	99,5	100	99,9	?Mitochondrial myopathy with lactic acidosis, 251950
PNPT1	96,9	86,1	100	99,9	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POLG	99,9	98,8	100	100	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	99,3	97,2	100	99,8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLR2A	100	100	100	100	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLRMT	85,5	65,9	100	100	No OMIM disease ID
PPA2	97,3	88,6	100	100	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPCS	100	99,1	100	100	Cardiomyopathy, dilated, 2C, 618189
PRKAA1	99,8	99,6	100	100	No OMIM disease ID
PRPS1	86,4	86,3	100	99,7	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070

					Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PTCD3	99,2	96,9	100	100	?Combined oxidative phosphorylation deficiency 51, 619057
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PUS1	99,9	98	99,9	98,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PYCR1	100	98,2	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,3	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYROXD1	92,1	78,7	100	100	Myopathy, myofibrillar, 8, 617258
QRSL1	98,6	92,8	100	99,9	Combined oxidative phosphorylation deficiency 40, 618835
RARS2	99,7	98,6	100	100	Pontocerebellar hypoplasia, type 6, 611523
RMND1	99,7	97,2	100	99,9	Combined oxidative phosphorylation deficiency 11, 614922
RNASEH1	98,7	95,8	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RRM1	99,9	99,5	100	100	No OMIM disease ID
RRM2B	100	99,8	100	99,9	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RTN4IP1	99,6	97,3	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RYR1	97,1	94	99,4	99	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320
SACS	99,9	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	98,5	97,9	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SARS2	95,7	94,5	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SCO1	97,6	94,4	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	99,9	97,9	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SDHA	84,5	77,9	100	100	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011

					Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SDHAF1	100	98,4	100	100	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	100	100	100	100	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHD	53,8	49	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SERAC1	99,6	99,5	100	99,9	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SFXN4	99,6	97,4	100	100	Combined oxidative phosphorylation deficiency 18, 615578
SLC19A2	100	98,5	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	97,8	97	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	96,9	89,8	99,7	98,2	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	76,4	70,3	100	100	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A12	100	99,2	100	100	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100	99,4	100	100	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A19	99,9	98	100	100	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A21	100	99,6	100	100	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	99,2	96,5	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99,3	98,8	99,7	99,7	Fontaine progeroid syndrome, 612289
SLC25A3	99,7	96,9	100	100	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100	100	100	99,9	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A38	97,4	93,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100	99,8	100	100	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184

SLC25A42	97,1	94,3	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,7	98,6	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SOD2	100	100	100	100	No OMIM disease ID
SPART	99,7	96,4	100	100	Troyer syndrome, 275900
SPATA5	99,8	99,5	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPG7	90,4	86,7	100	100	Spastic paraplegia 7, autosomal recessive, 607259
SQOR	100	98	100	100	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	99,8	97,8	100	100	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SSBP1	99,1	94,1	100	100	Optic atrophy 13 with retinal and foveal abnormalities, 165510
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
STAT2	100	99,4	100	100	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STXBP1	96,8	96,2	100	100	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	88,8	79,4	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100	99,7	100	99,8	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	91,7	79,1	100	100	No OMIM disease ID
SURF1	89,5	88,1	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SZT2	99,6	99,3	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TACO1	98,9	93,7	100	100	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAZ	99,3	93,7	100	100	Barth syndrome, 302060
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878

TAOK1	99,6	97,5	100	100	No OMIM disease ID
TARS2	99,9	98,8	100	100	?Combined oxidative phosphorylation deficiency 21, 615918
TDP2	99,6	99,5	100	99,9	Spinocerebellar ataxia, autosomal recessive 23, 616949
TFAM	98	78,5	100	100	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFB2M	100	98,5	100	99,9	No OMIM disease ID
THG1L	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 28, 618800
TIMM22	100	99,2	100	100	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100	99,9	100	100	No OMIM disease ID
TIMM50	98,4	95	100	100	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	96,2	83,1	100	100	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	99,9	99,8	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TK2	99	96	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TMEM126A	95,4	80	100	100	Optic atrophy 7, 612989
TMEM126B	99,6	97,2	100	100	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM186	100	100	100	100	No OMIM disease ID
TMEM65	89,3	83,2	95,1	87	No OMIM disease ID
TMEM70	98,4	94,6	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMX2	100	99,2	100	100	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TOMM70	99,9	99,3	100	100	No OMIM disease ID
TOP3A	99,6	96,5	100	100	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TPK1	99,5	97,2	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRIT1	100	100	100	100	Combined oxidative phosphorylation deficiency 35, 617873
TRMT10C	100	99,9	100	99,9	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	99,8	99,1	100	100	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	99,9	99,6	100	99,9	Liver failure, transient infantile, 613070
TRNT1	99,7	97,4	100	99,9	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959

TSFM	100	99,3	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TTC19	83,8	74,1	100	99,8	Mitochondrial complex III deficiency, nuclear type 2, 615157
TUFM	99,9	97,6	100	100	Combined oxidative phosphorylation deficiency 4, 610678
TWNK	100	99,9	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	100	98,5	100	100	?Combined oxidative phosphorylation deficiency 29, 616811
TYMP	100	99,4	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UQCC1	100	100	100	99,9	No OMIM disease ID
UQCC2	99,9	98,5	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100	97,5	100	100	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100	100	100	100	No OMIM disease ID
UQCR11	100	100	100	100	No OMIM disease ID
UQCRB	97,7	92,1	100	100	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	99,8	98,3	100	100	Parkinsonism with polyneuropathy, 619279
UQCRC2	100	98,8	100	100	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	94,1	88,8	100	100	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100	97,3	100	100	No OMIM disease ID
UQCRQ	100	100	100	100	Mitochondrial complex III deficiency, nuclear type 4, 615159
VARS2	100	99	100	100	Combined oxidative phosphorylation deficiency 20, 615917
VPS13D	100	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 4, 607317
WARS2	100	99,8	100	99,8	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WDR45	98,2	92	100	100	Neurodegeneration with brain iron accumulation 5, 300894
YARS2	99,9	99,4	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	98,9	93,7	100	100	?Optic atrophy 11, 617302

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.
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Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.

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 coverage denoting NC 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 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors