LIVER DISORDERS PANEL DG-5.1.0 (260 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100%	100%	100%	100%	99.7%	Achalasia-addisonianism-alacrimia syndrome, 231550
ABCB11	100%	100%	100%	100%	99.7%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	100%	100%	99.8%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	100%	100%	100%	99.8%	Dubin-Johnson syndrome, 237500
ABCD3	100%	100%	100%	100%	99.5%	?Bile acid synthesis defect, congenital, 5, 616278;Oculopharyngodistal myopathy 5, 621446
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACOX2	100%	100%	100%	100%	99.7%	Bile acid synthesis defect, congenital, 6, 617308
ACTA2	100%	100%	100%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042

ACTG2	93%	93%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431;Visceral myopathy 1, 155310
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADK	88%	88%	100%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	100%	100%	100%	99.9%	99.1%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1D1	88.4%	88.4%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
ALG8	78.4%	77.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.9%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type II, 608776
AMACR	100%	100%	100%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950

ANKS6	100%	99.9%	100%	100%	99.5%	Nephronophthisis 16, 615382
AP1S1	100%	100%	100%	100%	99.2%	MEDNIK syndrome, 609313
ARCN1	96.3%	96.1%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ARG1	93%	93%	100%	100%	99.8%	Argininemia, 207800
ASAH1	100%	100%	100%	99.9%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASS1	92.3%	90%	100%	100%	99.6%	Citrullinemia, 215700
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
ATP8B1	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300
B4GALT1	91%	88.3%	100%	100%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
BAAT	100%	100%	100%	100%	99.8%	Hypercholanemia, familial 3, 619232
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BLVRA	95.2%	95.2%	100%	100%	99.8%	Hyperbiliverdinemia, 614156

BOLA3	100%	100%	100%	100%	98.9%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
CARS1	100%	100%	100%	100%	99.6%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CASR	100%	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CDON	100%	100%	100%	100%	99.9%	Holoprosencephaly 11, 614226
CEL	100%	100%	99.9%	99.4%	97.7%	Maturity-onset diabetes of the young, type VIII, 609812
CELA3B	100%	100%	100%	99.8%	97.8%	
CEP83	100%	100%	100%	100%	99.8%	Nephronophthisis 18, 615862
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376

CFTR	100%	100%	100%	100%	99.6%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CHD8	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHRM3	100%	100%	100%	100%	99.8%	Prune belly syndrome, 100100
CHRNA3	92%	92%	100%	99.9%	99.4%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLDN1	100%	100%	100%	100%	99.9%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLMP	100%	100%	100%	100%	99.7%	Congenital short bowel syndrome, 615237
COG6	100%	100%	100%	100%	99.9%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	95.1%	95.1%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIe, 608779
COX4I2	100%	99.1%	100%	100%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714

CPA1	100%	100%	100%	100%	99.6%	
CTRC	100%	100%	100%	100%	99.6%	{Pancreatitis, chronic, susceptibility to}, 167800
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP7A1	100%	100%	100%	100%	99.8%	
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DGUOK	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900

DNAJB11	100%	100%	100%	100%	99.8%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJC21	94.7%	94.7%	100%	100%	99.3%	Bone marrow failure syndrome 3, 617052
DNASE2	92.7%	92.7%	100%	100%	99.3%	Autoinflammatory-panc ytopenia syndrome, 619858
EDN3	100%	100%	100%	99.9%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	100%	100%	100%	100%	99.5%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EFL1	96.3%	96.3%	100%	100%	99.9%	Shwachman-Diamond syndrome 2, 617941
EIF2AK3	100%	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
EPHX1	91.3%	90.8%	100%	100%	99.5%	
ERBB3	100%	100%	100%	99.9%	98.9%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleuke mia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680
FAH	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FARSB	94%	94%	100%	100%	99.7%	Rajab interstitial lung disease with brain calcifications 1, 613658

FECH	100%	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FLNA	100%	100%	98.3%	87.4%	68.7%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FOCAD	100%	100%	100%	99.9%	99.7%	Liver disease, severe congenital, 619991
FOXP3	100%	100%	99.1%	88.7%	71.1%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
GALE	100%	100%	100%	100%	99.6%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALK1	92.3%	92.3%	100%	100%	99.8%	Galactokinase deficiency with cataracts, 230200
GALM	84%	84%	100%	100%	99.7%	Galactosemia IV, 618881
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400

GANAB	100%	100%	100%	100%	99.6%	Polycystic kidney disease 3, 600666
GATA6	93.8%	93.8%	100%	99.8%	98.1%	Atrial septal defect 9, 614475;Persistent truncus arteriosus, 217095;Pancreatic agenesis and congenital heart defects, 600001;Atrioventricular septal defect 5, 614474;Tetralogy of Fallot, 187500
GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GDNF	100%	100%	100%	100%	99.3%	{Hirschsprung disease, susceptibility to, 3}, 613711
GFM1	96.6%	96.6%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 1, 609060

GLI3	99.3%	99.3%	100%	99.9%	99.2%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GLIS3	100%	100%	100%	100%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GPR161	100%	100%	100%	100%	99.5%	{Medulloblastoma predisposition syndrome}, 155255
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HAMP	100%	100%	100%	100%	99.9%	Hemochromatosis, type 2B, 613313
HFE	100%	99.7%	100%	100%	99.4%	Hemochromatosis, type 1, 235200
HMGCS2	97.5%	97.5%	100%	100%	99.2%	HMG-CoA synthase-2 deficiency, 605911
HNF1A	100%	100%	100%	100%	99.5%	Hepatic adenoma, somatic, 142330;Diabetes mellitus, insulin-dependent, 20, 612520;{Diabetes mellitus, noninsulin-dependent, 2}, 125853;MODY, type III, 600496;{Diabetes mellitus, insulin-dependent}, 222100;Renal cell carcinoma, 144700

HNF1B	100%	100%	100%	100%	99.6%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B7	100%	100%	100%	100%	99.5%	Bile acid synthesis defect, congenital, 1, 607765
IARS1	100%	100%	100%	100%	99.6%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IDS	100%	100%	99.4%	91.4%	72.9%	Mucopolysaccharidosis II, 309900
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866

IFT56	100%	100%	100%	100%	99.8%	Biliary, renal, neurologic, and skeletal syndrome, 619534
INSR	100%	100%	100%	100%	99.5%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
INVS	100%	100%	100%	100%	99.9%	Nephronophthisis 2, infantile, 602088
ITCH	92.6%	92.6%	100%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
JAG1	100%	100%	100%	100%	99.6%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
KCNMA1	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729
KIF12	100%	100%	100%	100%	99.3%	Cholestasis, progressive familial intrahepatic, 8, 619662
KIFBP	95.9%	95.9%	100%	99.9%	99.5%	Goldberg-Shprintzen megacolon syndrome, 609460

KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
L1CAM	100%	100%	98.4%	88.2%	69.2%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
LARS1	94.3%	94.3%	100%	100%	99.7%	?Infantile liver failure syndrome 1, 615438
LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIG3	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIPA	95.6%	95.2%	100%	100%	99.9%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LMOD1	100%	100%	100%	100%	98.8%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813

LRPPRC	96.8%	96.7%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LSR	83.5%	83.5%	100%	99.9%	99%	
LYN	100%	100%	100%	100%	99.3%	Autoinflammatory disease, systemic, with vasculitis, 620376
MAGEL2	100%	100%	100%	100%	99.7%	Schaaf-Yang syndrome, 615547
MARS1	89.3%	88.7%	100%	100%	99.5%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MED12	100%	99.9%	98.1%	87.5%	68.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MICOS13	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 37, 618329
MPI	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ib, 602579
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRPS23	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 46, 618952

MTM1	100%	100%	99.2%	92.2%	74.7%	Myopathy, centronuclear, X-linked, 310400
MVK	100%	100%	100%	100%	99.8%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYH11	100%	100%	100%	100%	99.3%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYL9	100%	100%	100%	100%	99.2%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365
MYLK	99.2%	99.2%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
MYO5B	100%	100%	100%	100%	99.5%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NHP2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 2, 613987

NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NOS1	100%	100%	100%	100%	99.2%	
NOTCH2	100%	100%	100%	100%	99.8%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPC1	100%	100%	100%	100%	99.7%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	99%	95.2%	100%	100%	99.6%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	100%	100%	99.8%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NR1H4	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic, 5, 617049
OTC	100%	100%	99.6%	91.8%	73.3%	Ornithine transcarbamylase deficiency, 311250
PCK1	100%	100%	100%	100%	99.7%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	100%	100%	99.5%	PEPCK deficiency, mitochondrial, 261650

PDX1	100%	100%	100%	100%	99.5%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	92.4%	92.4%	100%	100%	99.9%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	88.6%	88.6%	100%	100%	99.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100%	100%	100%	100%	99.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.8%	99%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHKG2	100%	100%	100%	100%	99.1%	Glycogen storage disease IXc, 613027

PHOX2B	100%	100%	100%	100%	98.6%	{Neuroblastoma, susceptibility to, 2}, 613013;Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880
PKD1	100%	99.9%	100%	99.9%	99.1%	Polycystic kidney disease 1, 173900
PKD1L1	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	100%	100%	100%	100%	99.1%	Polycystic kidney disease 2, 613095
PKHD1	100%	100%	100%	100%	99.7%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PNLIP	100%	100%	100%	100%	99.6%	?Pancreatic lipase deficiency, 614338
POLG	100%	100%	100%	100%	99.8%	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	95.8%	95.8%	100%	100%	99.5%	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
POMC	100%	100%	100%	100%	99.7%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
PRF1	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRKCSH	100%	99.6%	100%	99.9%	99.4%	Polycystic liver disease 1, 174050
PROKR2	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100%	100%	100%	99.9%	99.1%	Pituitary hormone deficiency, combined, 2, 262600
PRSS1	100%	100%	100%	99.6%	96.8%	Pancreatitis, hereditary, 167800
PSKH1	100%	100%	100%	100%	99.4%	Cholestasis, progressive familial intrahepatic, 13, 620962
PTF1A	100%	100%	100%	99.5%	97.6%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935

QRSL1	91.9%	89.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835
RAB27A	100%	100%	100%	100%	99.6%	Griscelli syndrome, type 2, 607624
RAD21	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RET	100%	100%	100%	100%	99.4%	{Hirschsprung disease, susceptibility to, 1}, 142623;Multiple endocrine neoplasia IIA, 171400;{Hirschsprung disease, protection against}, 142623;Medullary thyroid carcinoma, 155240;Pheochromocytoma, 171300;Multiple endocrine neoplasia IIB, 162300
RFX6	100%	100%	100%	100%	99.8%	Mitchell-Riley syndrome, 615710
RINT1	93.6%	93.6%	100%	100%	99.8%	Infantile liver failure syndrome 3, 618641
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
SBDS	100%	100%	100%	100%	99.9%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SC5D	100%	100%	100%	100%	99.9%	Lathosterolosis, 607330

SCO1	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC61B	100%	100%	100%	100%	99%	
SEC63	92.9%	92.9%	100%	100%	99.9%	Polycystic liver disease 2, 617004
SEMA7A	100%	100%	100%	100%	99.5%	?Cholestasis, progressive familial intrahepatic, 11, 619874;[Blood group, John-Milton-Hagen system], 614745
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	100%	100%	99.3%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490;Emphysema due to AAT deficiency, 613490;Emphysema-cirrhosis, due to AAT deficiency, 613490
SGO1	100%	100%	100%	100%	99.9%	Chronic atrial and intestinal dysrhythmia, 616201
SHH	100%	100%	100%	99.8%	98.4%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHPK	100%	100%	100%	100%	99.1%	[Sedoheptulokinase deficiency], 617213
SKIC3	98.9%	98.9%	100%	100%	99.9%	Trichohepatoenteric syndrome 1, 222470
SLC10A1	95.7%	95.7%	100%	100%	99.3%	Hypercholanemia, familial 2, 619256

SLC22A5	97.1%	97.1%	100%	100%	99.5%	Carnitine deficiency, systemic primary, 212140
SLC25A13	91.7%	91.7%	100%	100%	99.6%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	100%	100%	99.8%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC27A5	100%	100%	100%	100%	99.5%	
SLC37A4	100%	100%	100%	100%	99.8%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC40A1	100%	100%	100%	100%	99.9%	Hemochromatosis, type 4, 606069
SLC51A	96.2%	96.2%	100%	100%	99.6%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SLCO1B1	100%	100%	100%	99.9%	99.6%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100%	100%	100%	100%	99.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SMPD1	100%	100%	100%	100%	99.6%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200

SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPINK1	100%	100%	100%	100%	100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINT2	100%	100%	100%	100%	99.2%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPTB	100%	100%	100%	100%	99.4%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948;Spherocytosis, type 2, 616649
SRP54	96.3%	96.3%	100%	100%	99.9%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
STAT1	96%	96%	100%	100%	99.8%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162;Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STN1	87.1%	87.1%	100%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341

STX11	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX3	100%	100%	100%	100%	99.7%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445
STXBP2	100%	100%	100%	99.9%	99.3%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100%	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
TBX19	100%	100%	100%	100%	99.6%	Adrenocorticotropic hormone deficiency, 201400
TERC						Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550

TERT	99.6%	97.7%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TFR2	95.8%	94.2%	100%	100%	99%	Hemochromatosis, type 3, 604250
TGIF1	100%	100%	100%	100%	99.9%	Holoprosencephaly 4, 142946
TJP2	97.1%	95.8%	100%	100%	99.8%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TMEM70	100%	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMPRSS15	100%	100%	100%	100%	99.7%	Enterokinase deficiency, 226200
TNFAIP3	100%	100%	100%	100%	99.8%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185

TPO	100%	100%	100%	100%	99.5%	Thyroid dyshormonogenesis 2A, 274500
TRAF3IP1	100%	100%	100%	100%	99.6%	
TRMU	88%	88%	100%	100%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRPV6	100%	100%	100%	100%	99.2%	Hyperparathyroidism, transient neonatal, 618188
TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505
TULP3	93.4%	93.4%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TYMP	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	98%	97.9%	100%	100%	99.7%	Johanson-Blizzard syndrome, 243800
UGT1A1	100%	100%	100%	100%	99.6%	Crigler-Najjar syndrome, type I, 218800;[Bilirubin, serum level of, QTL1], 601816;Hyperbilirubinemia, familial transient neonatal, 237900;Crigler-Najjar syndrome, type II, 606785;[Gilbert syndrome], 143500

UNC13D	100%	100%	100%	100%	99.5%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	100%	100%	99.4%	Osteootohepatoenteric syndrome, 619377
UQCRC2	74.7%	74.7%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 5, 615160
USP53	100%	100%	100%	100%	99.8%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
UTP4	100%	100%	100%	100%	99.6%	
VIL1	100%	100%	100%	100%	99.1%	
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100%	100%	100%	100%	99.9%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS50	96.2%	95.3%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
WARS2	93.8%	93.8%	100%	100%	100%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710

WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectoderm dysplasia 2, 613610
XIAP	100%	100%	99.5%	93%	77.1%	Lymphoproliferative syndrome, X-linked, 2, 300635
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
ZEB2	100%	100%	100%	99.9%	99.4%	Mowat-Wilson syndrome, 235730
ZFYVE19	100%	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 9, 619849
ZNFX1	100%	100%	100%	100%	99.6%	Immunodeficiency 91 and hyperinflammation, 619644

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

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors