LIVER DISORDERS PANEL DG-4.1.0 (171 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*
ABCB11	100%	100%	100%	100%	99.6%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	100%	100%	99.7%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	100%	100%	100%	99.6%	Dubin-Johnson syndrome, 237500
ABCD3	100%	100%	100%	100%	99.8%	?Bile acid synthesis defect, congenital, 5, 616278
ACOX2	100%	100%	100%	100%	99.3%	Bile acid synthesis defect, congenital, 6, 617308

ACTA2	100%	100%	100%	100%	99.5%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTG2	100%	100%	100%	99.9%	99%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431;Visceral myopathy 1, 155310
ADK	90.9%	90.9%	100%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300
AHCY	100%	100%	100%	100%	99.3%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AKR1D1	100%	100%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555
ALDOB	100%	100%	100%	100%	99.6%	Fructose intolerance, hereditary, 229600
ALG8	77.9%	77.5%	100%	99.9%	99.5%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874

ALG9	100%	100%	100%	100%	99.6%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
AMACR	100%	100%	100%	100%	99.5%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
ANKS6	99.7%	98.6%	100%	100%	99.3%	Nephronophthisis 16, 615382
AP1S1	100%	100%	100%	99.9%	99.1%	MEDNIK syndrome, 609313
ARCN1	100%	100%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ATP7B	100%	100%	100%	100%	99.7%	Wilson disease, 277900
ATP8B1	100%	100%	100%	100%	99.5%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300

B4GALT1	100%	100%	100%	99.9%	98.7%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
BAAT	100%	100%	100%	100%	99.5%	Bile acid conjugation defect 1, 619232
BCS1L	100%	100%	100%	100%	99.4%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BLVRA	100%	100%	100%	100%	99.9%	Hyperbiliverdinemia, 614156
CARS1	100%	99.9%	100%	100%	99.2%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CC2D2A	98.2%	98.2%	100%	100%	99.6%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CEP83	100%	100%	100%	100%	99.7%	Nephronophthisis 18, 615862
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376

CFTR	100%	100%	100%	100%	99.5%	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%	99.4%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHRM3	100%	100%	100%	99.9%	98.4%	Prune belly syndrome, 100100
CHRNA3	100%	100%	100%	99.9%	99%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CLDN1	100%	100%	100%	100%	99.5%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLMP	100%	100%	100%	100%	99.4%	Congenital short bowel syndrome, 615237

COG6	100%	100%	100%	100%	99.7%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	100%	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type IIe, 608779
CYP27A1	100%	100%	100%	99.9%	99%	Cerebrotendinous xanthomatosis, 213700
CYP7B1	100%	100%	100%	100%	99.7%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
DCDC2	100%	100%	100%	100%	99.4%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394

DGUOK	100%	100%	100%	100%	99.8%	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%	96.1%	100%	100%	99.4%	Smith-Lemli-Opitz syndrome, 270400
DKC1	100%	99.4%	99%	89.6%	70.2%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNAJB11	100%	100%	100%	100%	99.4%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNASE2	100%	100%	100%	100%	99%	Autoinflammatory-pancytopenia syndrome, 619858

EDNRB	100%	100%	100%	100%	99.4%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EPHX1	100%	100%	100%	99.9%	98.7%	
ERBB3	100%	100%	100%	100%	98.8%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ETFDH	95.3%	92.7%	100%	100%	99.5%	Glutaric acidemia IIC, 231680
FAH	100%	100%	100%	100%	99.4%	Tyrosinemia, type I, 276700
FARSB	100%	100%	100%	100%	99.4%	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%	99.7%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812

FLNA	100%	99.8%	98.2%	86.8%	67.3%	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%	99.4%	Liver disease, severe congenital, 619991
GALM	100%	100%	100%	100%	99.3%	Galactosemia IV, 618881
GALT	100%	100%	100%	100%	99%	Galactosemia, 230400
GANAB	100%	100%	100%	100%	99.5%	Polycystic kidney disease 3, 600666

GBA1	100%	100%	100%	100%	99.1%	{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%	99.8%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GDNF	100%	100%	100%	100%	99%	{Hirschsprung disease, susceptibility to, 3}, 613711
GFM1	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 1, 609060

GLI3	99.3%	99.3%	100%	99.9%	99.1%	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%	99.1%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
HADHA	100%	100%	100%	100%	99.5%	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%	99.2%	Hemochromatosis, type 2B, 613313
HFE	100%	100%	100%	100%	98.7%	Hemochromatosis, type 1, 235200
HMGCS2	100%	100%	100%	99.9%	99.6%	HMG-CoA synthase-2 deficiency, 605911

HNF1B	100%	100%	100%	100%	99.4%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700
HSD17B4	100%	100%	100%	100%	99.6%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B7	100%	100%	100%	100%	99.6%	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%	99.3%	99.2%	91.1%	72.1%	Mucopolysaccharidosis II, 309900
IFT140	100%	100%	100%	100%	99%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781
IFT172	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630

IFT43	100%	100%	100%	100%	99.4%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT56	100%	100%	100%	100%	99.7%	Biliary, renal, neurologic, and skeletal syndrome, 619534
INSR	100%	100%	100%	99.9%	98.9%	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%	99.6%	Nephronophthisis 2, infantile, 602088
JAG1	100%	100%	100%	100%	99.4%	?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%	{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%	98.9%	Cholestasis, progressive familial intrahepatic, 8, 619662
L1CAM	100%	99.7%	98.4%	87.6%	68.6%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
LARS1	100%	100%	100%	99.9%	99.3%	?Infantile liver failure syndrome 1, 615438
LARS2	100%	100%	100%	100%	99.6%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LIG3	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780

LMOD1	100%	100%	100%	100%	98.2%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LRP5	100%	100%	100%	99.9%	98.3%	Osteopetrosis, autosomal dominant 1, 607634;[Bone mineral density variability 1], 601884;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Exudative vitreoretinopathy 4, 601813
LYN	100%	100%	100%	99.9%	99.6%	Autoinflammatory disease, systemic, with vasculitis, 620376
MAGEL2	100%	100%	100%	100%	99.4%	Schaaf-Yang syndrome, 615547
MARS1	100%	100%	100%	100%	99.2%	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%	99.6%	98.6%	87.4%	67.4%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MTM1	100%	99.9%	99.3%	92.2%	74.8%	Myopathy, centronuclear, X-linked, 310400
MYH11	100%	100%	100%	99.9%	98.7%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYL9	100%	100%	100%	99.9%	99.1%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365

MYLK	99.2%	99.2%	100%	99.9%	99.2%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
MYO5B	100%	100%	100%	100%	99.4%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
NBAS	100%	99.9%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NHP2	100%	100%	100%	99.9%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	92.5%	92.5%	100%	100%	98.8%	?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

NOTCH2	100%	100%	100%	100%	99.7%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NPC1	100%	100%	100%	99.9%	99.4%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	100%	100%	100%	99.9%	99.1%	Niemann-pick disease, type C2, 607625
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
PEX1	100%	100%	100%	100%	99.8%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871

PEX12	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	99.9%	98.4%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100%	100%	100%	100%	99.5%	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	100%	100%	100%	100%	99.3%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

PEX3	100%	100%	100%	100%	99.8%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.7%	98.5%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	99.8%	98.5%	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%	98.9%	Glycogen storage disease IXc, 613027
PKD1	99.9%	99.6%	100%	99.8%	98.3%	Polycystic kidney disease 1, 173900

PKD2	100%	100%	100%	99.8%	97.4%	Polycystic kidney disease 2, 613095
PKHD1	100%	100%	100%	99.9%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POLG	100%	100%	100%	100%	99.5%	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
POMC	100%	100%	100%	100%	99.5%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734

PRKCSH	100%	100%	100%	100%	99.2%	Polycystic liver disease 1, 174050
RAD21	100%	100%	100%	100%	99.8%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RFX6	100%	100%	100%	99.9%	99.5%	Mitchell-Riley syndrome, 615710
RINT1	100%	100%	100%	100%	99.6%	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
SC5D	100%	100%	100%	100%	99.8%	Lathosterolosis, 607330
SCO1	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCYL1	100%	100%	100%	99.9%	98.6%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SEC61B	100%	100%	100%	99.7%	98%	

SEC63	100%	100%	100%	100%	99.6%	Polycystic liver disease 2, 617004
SEMA7A	100%	100%	100%	99.9%	98.8%	?Cholestasis, progressive familial intrahepatic, 11, 619874;[Blood group, John-Milton-Hagen system], 614745
SERPINA1	100%	100%	100%	100%	98.4%	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%	99.7%	Chronic atrial and intestinal dysrhythmia, 616201
SKIC3	98.9%	98.9%	100%	100%	99.6%	Trichohepatoenteric syndrome 1, 222470
SLC10A1	100%	100%	100%	100%	99.1%	Hypercholanemia, familial 2, 619256
SLC25A13	100%	100%	100%	100%	99.7%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC40A1	100%	100%	100%	100%	99.7%	Hemochromatosis, type 4, 606069

SLC51A	100%	100%	100%	99.9%	98.4%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SMPD1	100%	100%	100%	99.9%	99.1%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SOX10	97.8%	97.8%	100%	99.9%	98.3%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPINT2	100%	100%	100%	99.9%	98.5%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
STN1	87.1%	87%	100%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TALDO1	100%	100%	100%	99.9%	97.6%	Transaldolase deficiency, 606003
TBX19	100%	100%	100%	99.9%	99.2%	Adrenocorticotropic hormone deficiency, 201400

TERC						Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100%	100%	100%	99.9%	98.4%	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	100%	100%	100%	99.9%	99%	Hemochromatosis, type 3, 604250
TJP2	100%	100%	100%	100%	99.6%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878

TMEM67	96.1%	96.1%	100%	100%	99.8%	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%	99.9%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TRAF3IP1	100%	100%	100%	100%	98.7%	Senior-Loken syndrome 9, 616629
TRMU	100%	100%	100%	99.9%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TULP3	100%	100%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902

TWNK	100%	100%	100%	100%	99.5%	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%	99.9%	98.8%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
UBR1	98%	98%	100%	100%	99.6%	Johanson-Blizzard syndrome, 243800
UGT1A1	100%	100%	100%	100%	99.3%	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%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	100%	100%	99.3%	Osteootohepatoenteric syndrome, 619377

USP53	100%	100%	100%	100%	99.5%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
VIPAS39	100%	100%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100%	100%	100%	99.9%	99.4%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS50	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
WDR35	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610

YARS1	100%	100%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
ZFYVE19	100%	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 9, 619849

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 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 4.1.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors