Oslo universitetssykehus
Avdeling for medisinsk genetikk

Medfødte leversykdommer

Genpanel, versjon v01

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet , gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner, så disse vil ikke bli analysert: **GBA, PRSS1, SBDS**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ABCB11	42	NM_003742.2		2-28	Cholestasis, progressive familial intrahepatic 2 OMIM Cholestasis, benign recurrent intrahepatic, 2 OMIM
ABCB4	45	NM_000443.3		2-28	Gallbladder disease 1 OMIM Cholestasis, progressive familial intrahepatic 3 OMIM Cholestasis, intrahepatic, of pregnancy, 3 OMIM
ABCC2	53	NM_000392.4		1-32	Dubin-Johnson syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ABCD3	67	NM_002858.3		1-23	?Bile acid synthesis defect, congenital, 5 OMIM
ABCG5	13886	NM_022436.2		1-13	Sitosterolemia OMIM
ABCG8	13887	NM_022437.2		1-13	Sitosterolemia OMIM
ACAD9	21497	NM_014049.4		1-18	Mitochondrial complex I deficiency due to ACAD9 deficiency OMIM
ACADM	89	NM_000016.5		1-12	Acyl-CoA dehydrogenase, medium chain, deficiency of OMIM
ACADVL	92	NM_000018.3		1-20	VLCAD deficiency OMIM
ADK	257	NM_001123.3		1-11	Hypermethioninemia due to adenosine kinase deficiency OMIM
AGL	321	NM_000642.2		2-34	Glycogen storage disease IIIb OMIM Glycogen storage disease IIIa OMIM
AHI1	21575	NM_017651.4		3-28	Joubert syndrome-3 OMIM
AKR1D1	388	NM_005989.3		1-9	Bile acid synthesis defect, congenital, 2 OMIM
ALAD	395	NM_000031.5		2-12	Porphyria, acute hepatic OMIM
ALDOA	414	NM_000034.3		7-14	Glycogen storage disease XII OMIM
ALDOB	417	NM_000035.3		2-9	Fructose intolerance OMIM
AMACR	451	NM_014324.5		1-5	Bile acid synthesis defect, congenital, 4 OMIM Alpha-methylacyl-CoA racemase deficiency OMIM
ANKS6	26724	NM_173551.3		1-15	Nephronophthisis 16 OMIM
AP1S1	559	NM_001283.3		1-5	MEDNIK syndrome OMIM

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ARL13B	25419	NM_182896.2		1-10	Joubert syndrome 8 OMIM
ARL6	13210	NM_177976.2		3-9	Bardet-Biedl syndrome 3 OMIM
ASL	746	NM_000048.3		2-17	Argininosuccinic aciduria OMIM
ASS1	758	NM_000050.4		3-16	Citrullinemia OMIM
ATP7B	870	NM_000053.3		1-21	Wilson disease OMIM
ATP8B1	3706	NM_005603.4		2-28	Cholestasis, progressive familial intrahepatic 1 OMIM Cholestasis, intrahepatic, of pregnancy, 1 OMIM Cholestasis, benign recurrent intrahepatic OMIM
B9D1	24123	NM_015681.3		1-7	Joubert syndrome 27 OMIM ?Meckel syndrome 9 OMIM
B9D2	28636	NM_030578.3		2-4	Meckel syndrome 10 OMIM
BAAT	932	NM_001701.3		2-4	Hypercholanemia, familial OMIM
BBS1	966	NM_024649.4		1-17	Bardet-Biedl syndrome 1 OMIM
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM
BBS12	26648	NM_152618.2		2	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3		1-17	Bardet-Biedl syndrome 2 OMIM Retinitis pigmentosa 74 OMIM
BBS4	969	NM_033028.4		1-16	Bardet-Biedl syndrome 4 OMIM
BBS5	970	NM_152384.2		1-12	Bardet-Biedl syndrome 5 OMIM

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BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
BCS1L	1020	NM_004328.4		3-9	Mitochondrial complex III deficiency, nuclear type 1 OMIM Leigh syndrome OMIM GRACILE syndrome OMIM Bjornstad syndrome OMIM
C10orf2	1160	NM_021830.4		1-5	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3 OMIM Perrault syndrome 5 OMIM Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) OMIM
C19orf70	33702	NM_205767.1		1-4	Mitochondrial encephalopathy with liver disease PubMed
C5orf42	25801	NM_023073.3		2-52	Joubert syndrome 17 OMIM Orofaciodigital syndrome VI OMIM
CC2D2A	29253	NM_001080522.2		3-38	Meckel syndrome 6 OMIM Joubert syndrome 9 OMIM COACH syndrome OMIM
CCDC115	28178	NM_032357.3		1-5	Congenital disorder of glycosylation, type IIo OMIM
CEP164	29182	NM_014956.4		3-33	Nephronophthisis 15 OMIM
CEP290	29021	NM_025114.3	54	2-54	Senior-Loken syndrome 6 OMIM Meckel syndrome 4 OMIM Leber congenital amaurosis 10 OMIM Joubert syndrome 5 OMIM ?Bardet-Biedl syndrome 14 OMIM
CEP41	12370	NM_018718.2		1-11	Joubert syndrome 15 OMIM
CFTR	1884	NM_000492.3		1-27	Cystic fibrosis OMIM {Pancreatitis, idiopathic} OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
CLDN1	2032	NM_021101.4		1-4	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis OMIM
COG1	6545	NM_018714.2		1-14	Congenital disorder of glycosylation, type IIg OMIM
COG6	18621	NM_020751.2		1-19	Congenital disorder of glycosylation, type III OMIM Shaheen syndrome OMIM
COG7	18622	NM_153603.3		1-17	Congenital disorder of glycosylation, type IIe OMIM
CPT1A	2328	NM_001876.3		2-19	CPT deficiency, hepatic, type IA OMIM
CPT2	2330	NM_000098.2		1-5	CPT II deficiency, myopathic, stress-induced OMIM CPT II deficiency, lethal neonatal OMIM CPT II deficiency, infantile OMIM
CYP27A1	2605	NM_000784.3		1-9	Cerebrotendinous xanthomatosis OMIM
CYP7B1	2652	NM_004820.3		1-6	Bile acid synthesis defect, congenital, 3 OMIM
DCDC2	18141	NM_016356.4		1-10	Nephronophthisis 19 OMIM
DGUOK	2858	NM_080916.2		1-7	Portal hypertension, noncirrhotic OMIM Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 OMIM
DHCR7	2860	NM_001360.2		3-9	Smith-Lemli-Opitz syndrome OMIM
DLD	2898	NM_000108.4		1-14	Dihydrolipoamide dehydrogenase deficiency OMIM
EARS2	29419	NM_001083614.1		1-9	Combined oxidative phosphorylation deficiency 12 OMIM

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EIF2AK3	3255	NM_004836.5		1-17	Wolcott-Rallison syndrome OMIM
EPHX1	3401	NM_000120.3		2-9	Hypercholanemia, familial OMIM
EPM2A	3413	NM_005670.3		1-4	Epilepsy, progressive myoclonic 2B (Lafora) OMIM Epilepsy, progressive myoclonic 2A (Lafora) OMIM
ETFA	3481	NM_000126.3		1-12	Glutaric acidemia IIA OMIM
ETFB	3482	NM_001985.2		1-6	Glutaric acidemia IIB OMIM
ETFDH	3483	NM_004453.3		1-13	Glutaric acidemia IIC OMIM
EVC	3497	NM_153717.2		1-21	Ellis-van Creveld syndrome OMIM Weyers acrodental dysostosis OMIM
EVC2	19747	NM_147127.4		1-22	Weyers acrofacial dysostosis OMIM Ellis-van Creveld syndrome OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FBP1	3606	NM_000507.3		1-7	Fructose-1,6-bisphosphatase deficiency OMIM
FH	3700	NM_000143.3		1-10	Fumarase deficiency OMIM
G6PC	4056	NM_000151.3		1-5	Glycogen storage disease Ia OMIM
GAA	4065	NM_000152.3		2-20	Glycogen storage disease II OMIM
GALE	4116	NM_000403.3		3-12	Galactose epimerase deficiency OMIM
GALK1	4118	NM_000154.1		1-8	Galactokinase deficiency with cataracts OMIM
GALT	4135	NM_000155.3		1-11	Galactosemia OMIM

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GANAB	4138	NM_198335.3		1-25	Polycyctic kidney disease 3 OMIM
GBA	4177	NM_001005741.2	2-12	2-12	Gaucher disease, type II OMIM Gaucher disease, type I OMIM Gaucher disease, perinatal lethal OMIM Gaucher disease, type IIIC OMIM Gaucher disease, type III OMIM
GBE1	4180	NM_000158.3		1-16	Polyglucosan body disease, adult form OMIM Glycogen storage disease IV OMIM
GFM1	13780	NM_024996.5		1-18	Combined oxidative phosphorylation deficiency 1 OMIM
GLIS2	29450	NM_032575.2		1-6	Nephronophthisis 7 OMIM
GYS2	4707	NM_021957.3		1-16	Glycogen storage disease 0, liver OMIM
HADHA	4801	NM_000182.4		1-20	LCHAD deficiency OMIM Trifunctional protein deficiency OMIM
HADHB	4803	NM_000183.2		2-16	Trifunctional protein deficiency OMIM
HMGCL	5005	NM_000191.2		1-9	HMG-CoA lyase deficiency OMIM
HNF1A	11621	NM_000545.5		1-10	Neonatal cholestasis and MODY OMIM PubMed
HNF1B	11630	NM_000458.3		1-9	Neonatal cholestasis, renal cysts and diabetes syndrome OMIM
HSD17B4	5213	NM_000414.3		1-24	Perrault syndrome 1 OMIM D-bifunctional protein deficiency OMIM
HSD3B7	18324	NM_025193.3		2-7	Bile acid synthesis defect, congenital, 1 OMIM
IARS	5330	NM_002161.5		2-34	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
IFT140	29077	NM_014714.3		3-31	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT43	29669	NM_052873.2		1-8	Cranioectodermal dysplasia 3 OMIM
IFT80	29262	NM_020800.2		2-20	Short-rib thoracic dysplasia 2 with or without polydactyly OMIM
INPP5E	21474	NM_019892.4		1-10	Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM Joubert syndrome 1 OMIM
INSR	6091	NM_000208.2		1-22	Rabson-Mendenhall syndrome OMIM Leprechaunism/Donohue syndrome OMIM
INVS	17870	NM_014425.3		2-17	Nephronophthisis 2, infantile OMIM
IQCB1	28949	NM_001023570.2		3-15	Senior-Loken syndrome 5 OMIM
JAG1	6188	NM_000214.2		1-26	Alagille syndrome 1 OMIM
KIF7	30497	NM_198525.2		2-19	Joubert syndrome 12 OMIM Acrocallosal syndrome OMIM
KRT18	6430	NM_000224.2		1-7	Cirrhosis, cryptogenic OMIM
KRT8	6446	NM_002273.3		1-8	Cirrhosis, cryptogenic OMIM
LARS	6512	NM_020117.9		1-32	Infantile liver failure syndrome 1 OMIM
LBR	6518	NM_002296.3		2-14	Reynolds syndrome OMIM Pelger-Huet anomaly OMIM Greenberg skeletal dysplasia OMIM
LIPA	6617	NM_000235.3		2-10	Wolman disease OMIM Cholesteryl ester storage disease OMIM

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LRP5	6697	NM_002335.3	1, 3-9	1-23	Hyperostosis, endosteal OMIM van Buchem disease, type 2 OMIM Osteosclerosis OMIM Osteoporosis-pseudoglioma syndrome OMIM Osteopetrosis, autosomal dominant 1 OMIM
MCEE	16732	NM_032601.3		1-3	Methylmalonyl-CoA epimerase deficiency OMIM
MKKS	7108	NM_018848.3		3-6	McKusick-Kaufman syndrome OMIM Bardet-Biedl syndrome 6 OMIM
MKS1	7121	NM_017777.3		1-18	Meckel syndrome 1 OMIM Joubert syndrome 28 OMIM Bardet-Biedl syndrome 13 OMIM
MMAA	18871	NM_172250.2		2-7	Methylmalonic aciduria, vitamin B12-responsive OMIM
MMAB	19331	NM_052845.3		1-9	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type OMIM
MMADHC	25221	NM_015702.2		2-8	Methylmalonic aciduria, cblD type, variant 2 OMIM Methylmalonic aciduria and homocystinuria, cblD type OMIM Homocystinuria, cblD type, variant 1 OMIM
MPI	7216	NM_002435.2		1-8	Congenital disorder of glycosylation, type Ib OMIM
MPV17	7224	NM_002437.4		2-8	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type) OMIM
MUT	7526	NM_000255.3		2-13	Methylmalonic aciduria, mut(0) type OMIM
MYO5B	7603	NM_001080467.2	40	1-40	Microvillus inclusion disease OMIM
NARS2	26274	NM_024678.5		1-14	Combined oxidative phosphorylation deficiency 24 OMIM

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NBAS	15625	NM_015909.3		1-52	Short stature, optic nerve atrophy, and Pelger-Huet anomaly OMIM Infantile liver failure syndrome 2 OMIM
NEK8	13387	NM_178170.2		1-15	Renal-hepatic-pancreatic dysplasia 2 OMIM
NHLRC1	21576	NM_198586.2		1	Epilepsy, progressive myoclonic 2B (Lafora) OMIM
NOTCH2	7882	NM_024408.3	1-4	1-34	Hajdu-Cheney syndrome OMIM Alagille syndrome 2 OMIM
NPC1	7897	NM_000271.4		1-25	Niemann-Pick disease, type C1 OMIM Niemann-Pick disease, type D OMIM
NPC2	14537	NM_006432.3		1-5	Niemann-pick disease, type C2 OMIM
NPHP1	7905	NM_000272.3		1-20	Nephronophthisis 1, juvenile OMIM Joubert syndrome 4 OMIM Senior-Loken syndrome-1 OMIM
NPHP3	7907	NM_153240.4		1-27	Renal-hepatic-pancreatic dysplasia 1 OMIM Nephronophthisis 3 OMIM Meckel syndrome 7 OMIM
NPHP4	19104	NM_015102.4		2-30	Senior-Loken syndrome 4 OMIM Nephronophthisis 4 OMIM
NR1H4	7967	NM_005123.3		3-11	Cholestasis, progressive familial intrahepatic, 5 OMIM
OFD1	2567	NM_003611.2		1-23	Simpson-Golabi-Behmel syndrome, type 2 OMIM Orofaciodigital syndrome I OMIM Joubert syndrome 10 OMIM
PARS2	30563	NM_152268.3		2	Alpers syndrome PubMed
PCCA	8653	NM_000282.3		1-24	Propionicacidemia OMIM
PCCB	8654	NM_000532.4		1-15	Propionicacidemia OMIM

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PEX1	8850	NM_000466.2		1-24	Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM Peroxisome biogenesis disorder 1A (Zellweger) OMIM Heimler syndrome 1 OMIM
PEX10	8851	NM_153818.1		1-6	Peroxisome biogenesis disorder 6B OMIM Peroxisome biogenesis disorder 6A (Zellweger) OMIM
PEX11B	8853	NM_003846.2		1-4	Peroxisome biogenesis disorder 14B OMIM
PEX12	8854	NM_000286.2		1-3	Peroxisome biogenesis disorder 3B OMIM Peroxisome biogenesis disorder 3A (Zellweger) OMIM
PEX13	8855	NM_002618.3		1-4	Peroxisome biogenesis disorder 11A (Zellweger) OMIM Peroxisome biogenesis disorder 11B OMIM
PEX14	8856	NM_004565.2		1-9	Peroxisome biogenesis disorder 13A (Zellweger) OMIM
PEX16	8857	NM_004813.2		1-11	Peroxisome biogenesis disorder 8B OMIM Peroxisome biogenesis disorder 8A (Zellweger) OMIM
PEX19	9713	NM_002857.3		1-8	Peroxisome biogenesis disorder 12A (Zellweger) OMIM
PEX2	9717	NM_000318.2		4	Peroxisome biogenesis disorder 5B OMIM Peroxisome biogenesis disorder 5A (Zellweger) OMIM
PEX26	22965	NM_017929.5		2-6	Peroxisome biogenesis disorder 7B OMIM Peroxisome biogenesis disorder 7A (Zellweger) OMIM
PEX3	8858	NM_003630.2		1-12	Peroxisome biogenesis disorder 10A (Zellweger) OMIM

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PEX5	9719	NM_001131025.1		2-16	Rhizomelic chondrodysplasia punctata, type 5 OMIM Peroxisome biogenesis disorder 2B OMIM Peroxisome biogenesis disorder 2A (Zellweger) OMIM
PEX6	8859	NM_000287.3		1-17	Peroxisome biogenesis disorder 4B OMIM Peroxisome biogenesis disorder 4A (Zellweger) OMIM Heimler syndrome 2 OMIM
PEX7	8860	NM_000288.3		1-10	Peroxisome biogenesis disorder 9B OMIM Rhizomelic chondrodysplasia punctata, type 1 OMIM
PGM1	8905	NM_002633.2		1-11	Congenital disorder of glycosylation, type It OMIM
PHKA2	8926	NM_000292.2		1-33	Glycogen storage disease, type IXa1 OMIM Glycogen storage disease, type IXa2 OMIM
PHKB	8927	NM_000293.2		1-31	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive OMIM
PHKG2	8931	NM_000294.2		2-10	Glycogen storage disease IXc OMIM Cirrhosis due to liver phosphorylase kinase deficiency
PKD2	9009	NM_000297.3		1-15	Polycystic kidney disease 2 OMIM
PKHD1	9016	NM_138694.3		2-67	Polycystic kidney and hepatic disease OMIM
PKLR	9020	NM_000298.5		1-11	Pyruvate kinase deficiency OMIM Adenosine triphosphate, elevated, of erythrocytes OMIM
PMM2	9115	NM_000303.2		1-8	Congenital disorder of glycosylation, type Ia OMIM

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POLG	9179	NM_002693.2		2-23	Mitochondrial DNA depletion syndrome 4B (MNGIE type) OMIM Mitochondrial DNA depletion syndrome 4A (Alpers type) OMIM Progressive external ophthalmoplegia, autosomal recessive 1 OMIM Progressive external ophthalmoplegia, autosomal dominant 1 OMIM Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) OMIM
POLG2	9180	NM_007215.3		1-8	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4 OMIM
POU1F1	9210	NM_000306.3		1-6	Pituitary hormone deficiency, combined, 1 OMIM
PRKCSH	9411	NM_002743.3		2-17	Polycystic liver disease 1 OMIM
PRSS1	9475	NM_002769.4	1-5	1-5	Trypsinogen deficiency OMIM Pancreatitis, hereditary OMIM
PYGL	9725	NM_002863.4		1-20	Glycogen storage disease VI OMIM
RPGRIP1L	29168	NM_015272.2		2-27	Joubert syndrome 7 OMIM COACH syndrome OMIM Meckel syndrome 5 OMIM
RRM2B	17296	NM_015713.4		1-9	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5 OMIM Mitochondrial DNA depletion syndrome 8B (MNGIE type) OMIM Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) OMIM
SBDS	19440	NM_016038.2	1-5	1-5	Shwachman-Diamond syndrome OMIM
SC5D	10547	NM_006918.4		2-5	Lathosterolosis OMIM

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SEC63	21082	NM_007214.4		1-21	Polycystic liver disease 2 OMIM
SERPINA1	8941	NM_000295.4		2-5	Hemorrhagic diathesis due to antithrombin Pittsburgh OMIM Emphysema-cirrhosis, due to AAT deficiency OMIM Emphysema due to AAT deficiency OMIM
SLC10A2	10906	NM_000452.2		1-6	Bile acid malabsorption, primary OMIM
SLC17A5	10933	NM_012434.4		1-11	Salla disease OMIM Sialic acid storage disorder, infantile OMIM
SLC22A5	10969	NM_003060.3		1-10	Carnitine deficiency, systemic primary OMIM
SLC25A13	10983	NM_014251.2		1-18	Citrullinemia, type II, neonatal-onset OMIM Citrullinemia, adult-onset type II OMIM
SLC25A20	1421	NM_000387.5		1-9	Carnitine-acylcarnitine translocase deficiency OMIM
SLC37A4	4061	NM_001164277.1		3-11	Glycogen storage disease Ib OMIM Glycogen storage disease Ic OMIM
SMPD1	11120	NM_000543.4		1-6	Niemann-Pick disease, type B OMIM Niemann-Pick disease, type A OMIM
SPINK1	11244	NM_003122.4		2-5	Tropical calcific pancreatitis OMIM Pancreatitis, hereditary OMIM
SUCLA2	11448	NM_003850.2		1-11	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) OMIM
SUCLG1	11449	NM_003849.3		1-9	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) OMIM
TCTN1	26113	NM_001082538.2		1-14	Joubert syndrome 13 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
TCTN2	25774	NM_024809.4		1-18	Joubert syndrome 24 OMIM ?Meckel syndrome 8 OMIM
TCTN3	24519	NM_015631.5		1-14	Orofaciodigital syndrome IV OMIM Joubert syndrome 18 OMIM
TFAM	11741	NM_003201.2	7	1-7	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type) OMIM
TJP2	11828	NM_004817.3		1-23	Cholestasis, progressive familial intrahepatic 4 OMIM Hypercholanemia, familial OMIM
TMEM138	26944	NM_016464.4		2-5	Joubert syndrome 16 OMIM
TMEM216	25018	NM_001173990.2		1-5	Joubert syndrome 2 OMIM Meckel syndrome 2 OMIM
TMEM231	37234	NM_001077416.2		1-6	Meckel syndrome 11 OMIM Joubert syndrome 20 OMIM
TMEM237	14432	NM_001044385.2		1-12	Joubert syndrome 14 OMIM
TMEM67	28396	NM_153704.5		1-28	Joubert syndrome 6 OMIM COACH syndrome OMIM Nephronophthisis 11 OMIM Meckel syndrome 3 OMIM
TMEM70	26050	NM_017866.5		1-3	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2 OMIM
TRIM32	16380	NM_012210.3		2	Muscular dystrophy, limb-girdle, type 2H OMIM ?Bardet-Biedl syndrome 11 OMIM
TRMU	25481	NM_018006.4		1-11	Liver failure, transient infantile OMIM
TTC21B	25660	NM_024753.4		1-29	Short-rib thoracic dysplasia 4 with or without polydactyly OMIM Nephronophthisis 12 OMIM
TTC37	23639	NM_014639.3		4-43	Trichohepatoenteric syndrome 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
TTC8	20087	NM_198309.3		2-15	Bardet-Biedl syndrome 8 OMIM
TYMP	3148	NM_001953.4		2-10	Mitochondrial DNA depletion syndrome 1 (MNGIE type) OMIM
UBR1	16808	NM_174916.2		1-47	Johanson-Blizzard syndrome OMIM
UGT1A1	12530	NM_000463.2		1-5	Crigler-Najjar syndrome, type II OMIM Crigler-Najjar syndrome, type I OMIM Hyperbilirubinemia, familial transient neonatal OMIM
VIPAS39	20347	NM_022067.3		3-21	Arthrogryposis, renal dysfunction, and cholestasis 2 OMIM
VPS33B	12712	NM_018668.4		1-23	Arthrogryposis, renal dysfunction, and cholestasis 1 OMIM
WDR19	18340	NM_025132.3		1-36	?Short-rib thoracic dysplasia 5 with or without polydactyly OMIM ?Cranioectodermal dysplasia 4 OMIM Senior-Loken syndrome 8 OMIM Nephronophthisis 13 OMIM
ZNF423	16762	NM_015069.3		1-8	Joubert syndrome 19 OMIM Nephronophthisis 14 OMIM