Ciliopati

Genpanel, versjon v05

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Sangersekvensering for påvisning av den sykdomsgivende varianten i CEP290 c.2991+1655A>G er utført
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
ACVR2B	174	NM_001106.3	100%	Heterotaxy, visceral, 4, autosomal OMIM
AHI1	21575	NM_017651.4	100%	Joubert syndrome-3 OMIM
AIPL1	359	NM_014336.4	100%	Retinitis pigmentosa, juvenile OMIM
ALMS1	428	NM_015120.4	99%	Alstrom syndrome OMIM
ANKS6	26724	NM_173551.3	96%	Nephronophthisis 16 OMIM
ARL13B	25419	NM_182896.2	100%	Joubert syndrome 8 OMIM
ARL6	13210	NM_177976.2	100%	{Bardet-Biedl syndrome 1, modifier of} OMIM Bardet-Biedl syndrome 3 OMIM
ARMC4	25583	NM_018076.3	94%	Ciliary dyskinesia, primary, 23 OMIM
ATXN10	10549	NM_013236.3	100%	Spinocerebellar ataxia 10 OMIM
B9D1	24123	NM_015681.3	100%	Joubert syndrome 27 OMIM
B9D2	28636	NM_030578.3	100%	Meckel syndrome 10 OMIM
BBIP1	28093	NM_001195306.1	100%	Bardet-Biedl syndrome 18 OMIM
BBS1	966	NM_024649.4	100%	Bardet-Biedl syndrome 1 OMIM
BBS10	26291	NM_024685.3	100%	Bardet-Biedl syndrome 10 OMIM

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BBS12	26648	NM_152618.2	100%	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3	100%	Retinitis pigmentosa 74 OMIM Bardet-Biedl syndrome 2 OMIM
BBS4	969	NM_033028.4	100%	Bardet-Biedl syndrome 4 OMIM
BBS5	970	NM_152384.2	99%	Bardet-Biedl syndrome 5 OMIM
BBS7	18758	NM_176824.2	99%	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2	99%	Bardet-Biedl syndrome 9 OMIM
C21orf59	1301	NM_021254.2	100%	Ciliary dyskinesia, primary, 26 OMIM
C2CD3	24564	NM_015531.5	100%	Orofaciodigital syndrome XIV OMIM
C2orf71	34383	NM_001029883.2	99%	Retinitis pigmentosa 54 OMIM
C5orf42	25801	NM_023073.3	99%	Orofaciodigital syndrome VI OMIM Joubert syndrome 17 OMIM
CABP4	1386	NM_145200.3	100%	Cone-rod synaptic disorder, congenital nonprogressive OMIM
CC2D2A	29253	NM_001080522.2	100%	Meckel syndrome 6 OMIM Joubert syndrome 9 OMIM COACH syndrome OMIM
CCDC103	32700	NM_213607.2	100%	Ciliary dyskinesia, primary, 17 OMIM
CCDC11	26530	NM_145020.3	99%	Heterotaxy, visceral, 6, autosomal recessive OMIM
CCDC114	26560	NM_144577.3	100%	Ciliary dyskinesia, primary, 20 OMIM
CCDC151	28303	NM_145045.4	100%	Ciliary dyskinesia, primary, 30 OMIM
CCDC28B	28163	NM_024296.4	100%	{Bardet-Biedl syndrome 1, modifier of} OMIM
CCDC39	25244	NM_181426.1	99%	Ciliary dyskinesia, primary, 14 OMIM

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CCDC40	26090	NM_017950.3	99%	Ciliary dyskinesia, primary, 15 OMIM
CCDC41	17966	NM_016122.2	99%	Nephronophthisis 18 OMIM
CCDC65	29937	NM_033124.4	100%	Ciliary dyskinesia, primary, 27 OMIM
CCNO	18576	NM_021147.4	99%	Ciliary dyskinesia, primary, 29 OMIM
CDH23	13733	NM_022124.5	100%	Usher syndrome, type 1D/F digenic OMIM Usher syndrome, type 1D OMIM
CEP104	24866	NM_014704.3	100%	Joubert syndrome 25 OMIM
CEP120	26690	NM_153223.3	100%	Short-rib thoracic dysplasia 13 with or without polydactyly OMIM
CEP164	29182	NM_014956.4	99%	Nephronophthisis 15 OMIM
CEP290	29021	NM_025114.3	98%	Leber congenital amaurosis 10 OMIM Joubert syndrome 5 OMIM ?Bardet-Biedl syndrome 14 OMIM Senior-Loken syndrome 6 OMIM Meckel syndrome 4 OMIM
CEP41	12370	NM_018718.2	99%	Joubert syndrome 15 OMIM
CLRN1	12605	NM_174878.2	100%	Usher syndrome, type 3A OMIM Retinitis pigmentosa 61 OMIM
CRB1	2343	NM_201253.2	100%	Retinitis pigmentosa-12, autosomal recessive OMIM Pigmented paravenous chorioretinal atrophy OMIM Leber congenital amaurosis 8 OMIM
CRELD1	14630	NM_015513.4	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome OMIM
CRX	2383	NM_000554.4	100%	Cone-rod retinal dystrophy-2 OMIM Leber congenital amaurosis 7 OMIM
CSPP1	26193	NM_024790.6	100%	Joubert syndrome 21 OMIM
DCDC2	18141	NM_016356.4	100%	Nephronophthisis 19 OMIM ?Deafness, autosomal recessive 66 OMIM
DDX59	25360	NM_001031725.4	100%	Orofaciodigital syndrome V OMIM

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DNAAF1	30539	NM_178452.4	100%	Ciliary dyskinesia, primary, 13 OMIM
DNAAF2	20188	NM_018139.2	100%	Ciliary dyskinesia, primary, 10 OMIM
DNAAF3	30492	NM_001256714.1	99%	Ciliary dyskinesia, primary, 2 OMIM
DNAH11	2942	NM_001277115.1	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus OMIM
DNAH5	2950	NM_001369.2	99%	Ciliary dyskinesia, primary, 3, with or without situs inversus OMIM
DNAI1	2954	NM_012144.3	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus OMIM
DNAI2	18744	NM_023036.4	99%	Ciliary dyskinesia, primary, 9, with or without situs inversus OMIM
DNAJB13	30718	NM_153614.3	100%	Ciliary dyskinesia, primary, 34 OMIM
DNAL1	23247	NM_031427.3	100%	Ciliary dyskinesia, primary, 16 OMIM
DPH1	3003	NM_001383.3	100%	Developmental delay with short stature, dysmorphic features, and sparse hair OMIM
DRC1	24245	NM_145038.3	100%	Ciliary dyskinesia, primary, 21 OMIM
DYNC2H1	2962	NM_001080463.1	99%	Short-rib thoracic dysplasia 3 with or without polydactyly OMIM
DYNC2LI1	24595	NM_001193464.1	100%	Short-rib throacic dysplasia 15 with polydactyly OMIM
DYX1C1	21493	NM_130810.3	100%	Ciliary dyskinesia, primary, 25 OMIM
EVC	3497	NM_153717.2	96%	Ellis-van Creveld syndrome OMIM Weyers acrodental dysostosis OMIM
EVC2	19747	NM_147127.4	98%	Weyers acrofacial dysostosis OMIM Ellis-van Creveld syndrome OMIM
GAS8	4166	NM_001286209.1	99%	Ciliary dyskinesia, primary, 33 OMIM
GDF6	4221	NM_001001557.2	100%	Leber congenital amaurosis 17 OMIM Klippel-Feil syndrome 1, autosomal dominant OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
GLI3	4319	NM_000168.5	100%	Greig cephalopolysyndactyly syndrome OMIM Polydactyly, preaxial, type IV OMIM Polydactyly, postaxial, types A1 and B OMIM Pallister-Hall syndrome OMIM
GLIS2	29450	NM_032575.2	100%	Nephronophthisis 7 OMIM
GPR98	17416	NM_032119.3	99%	Usher syndrome, type 2C OMIM Usher syndrome, type 2C, GPR98/PDZD7 digenic OMIM
GUCY2D	4689	NM_000180.3	100%	Leber congenital amaurosis 1 OMIM Cone-rod dystrophy 6 OMIM
HEATR2	26013	NM_017802.3	95%	Ciliary dyskinesia, primary, 18 OMIM
HNF1B	11630	NM_000458.3	99%	Renal cysts and diabetes syndrome OMIM
HYLS1	26558	NM_145014.2	100%	Hydrolethalus syndrome OMIM
IFT122	13556	NM_052985.3	100%	Cranioectodermal dysplasia 1 OMIM
IFT140	29077	NM_014714.3	99%	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT172	30391	NM_015662.2	99%	Short-rib thoracic dysplasia 10 with or without polydactyly OMIM Retinitis pigmentosa 71 OMIM
IFT27	18626	NM_006860.4	100%	?Bardet-Biedl syndrome 19 OMIM
IFT43	29669	NM_052873.2	100%	Cranioectodermal dysplasia 3 OMIM
IFT52	15901	NM_016004.3	100%	Short-rib thoracic dysplasia 16 with or without polydactyly OMIM
IFT74	21424	NM_001099223.1	99%	?Bardet-Biedl syndrome 20 OMIM
IFT80	29262	NM_020800.2	99%	Short-rib thoracic dysplasia 2 with or without polydactyly OMIM
IMPDH1	6052	NM_000883.3	97%	Retinitis pigmentosa 10 OMIM Leber congenital amaurosis 11 OMIM
INPP5E	21474	NM_019892.4	99%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM Joubert syndrome 1 OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
INVS	17870	NM_014425.3	100%	Nephronophthisis 2, infantile OMIM
IQCB1	28949	NM_001023570.2	97%	Senior-Loken syndrome 5 OMIM
KCNJ13	6259	NM_002242.4	100%	Leber congenital amaurosis 16 OMIM
KIAA0556	29068	NM_015202.2	99%	Joubert syndrome 26 OMIM
KIAA0586	19960	NM_001244192.1	99%	Short-rib thoracic dysplasia 14 with polydactyly OMIM Joubert syndrome 23 OMIM
KIF7	30497	NM_198525.2	97%	Joubert syndrome 12 OMIM Acrocallosal syndrome OMIM
LBR	6518	NM_002296.3	99%	Pelger-Huet anomaly OMIM
LCA5	31923	NM_181714.3	100%	Leber congenital amaurosis 5 OMIM
LRAT	6685	NM_004744.4	100%	Retinitis pigmentosa, juvenile OMIM Retinal dystrophy, early-onset severe OMIM Leber congenital amaurosis 14 OMIM
LRRC6	16725	NM_012472.4	99%	Ciliary dyskinesia, primary, 19 OMIM
LZTFL1	6741	NM_020347.3	100%	Bardet-Biedl syndrome 17 OMIM
MAPKBP1	29536	NM_014994.2	100%	Nephronophthisis 20 OMIM
MCIDAS	40050	NM_001190787.1	99%	Mucociliary clearance disorder
MKKS	7108	NM_018848.3	100%	McKusick-Kaufman syndrome OMIM Bardet-Biedl syndrome 6 OMIM
MKS1	7121	NM_017777.3	100%	Bardet-Biedl syndrome 13 OMIM Meckel syndrome 1 OMIM Joubert syndrome 28 OMIM
MUC1	7508	NM_002456.5	100%	Medullary cystic kidney disease 1 OMIM
MYO7A	7606	NM_000260.3	99%	Usher syndrome, type 1B OMIM
NEK1	7744	NM_012224.2	100%	Short-rib thoracic dysplasia 6 with or without polydactyly OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
NEK8	13387	NM_178170.2	100%	?Renal-hepatic-pancreatic dysplasia 2 OMIM ?Nephronophthisis 9 OMIM
NME8	16473	NM_016616.4	99%	Ciliary dyskinesia, primary, 6 OMIM
NMNAT1	17877	NM_022787.3	100%	Leber congenital amaurosis 9 OMIM
NODAL	7865	NM_018055.4	100%	Heterotaxy, visceral, 5 OMIM
NPHP1	7905	NM_000272.3	100%	Nephronophthisis 1, juvenile OMIM Joubert syndrome 4 OMIM Senior-Loken syndrome-1 OMIM
NPHP3	7907	NM_153240.4	99%	Renal-hepatic-pancreatic dysplasia 1 OMIM Nephronophthisis 3 OMIM Meckel syndrome 7 OMIM
NPHP4	19104	NM_015102.4	99%	Senior-Loken syndrome 4 OMIM Nephronophthisis 4 OMIM
OFD1	2567	NM_003611.2	94%	Orofaciodigital syndrome I OMIM Joubert syndrome 10 OMIM ?Retinitis pigmentosa 23 OMIM Simpson-Golabi-Behmel syndrome, type 2 OMIM
PCDH15	14674	NM_033056.3	100%	Usher syndrome, type 1F OMIM Usher syndrome, type 1D/F digenic OMIM
PDE6D	8788	NM_002601.3	100%	?Joubert syndrome 22 OMIM
PKHD1	9016	NM_138694.3	100%	Polycystic kidney and hepatic disease OMIM
PRPH2	9942	NM_000322.4	100%	Leber congenital amaurosis 18 OMIM Retinitis pigmentosa 7 and digenic OMIM
RD3	19689	NM_183059.2	100%	Leber congenital amaurosis 12 OMIM
RDH12	19977	NM_152443.2	100%	Leber congenital amaurosis 13 OMIM
REN	9958	NM_000537.3	100%	Renal tubular dysgenesis OMIM
RPE65	10294	NM_000329.2	100%	Retinitis pigmentosa 20 OMIM Leber congenital amaurosis 2 OMIM
RPGR	10295	NM_000328.2	99%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness OMIM Retinitis pigmentosa 3 OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
RPGRIP1	13436	NM_020366.3	100%	Leber congenital amaurosis 6 OMIM
RPGRIP1L	29168	NM_015272.2	96%	Meckel syndrome 5 OMIM Joubert syndrome 7 OMIM COACH syndrome OMIM
RSPH1	12371	NM_080860.3	100%	Ciliary dyskinesia, primary, 24 OMIM
RSPH3	21054	NM_031924.4	100%	Ciliary dyskinesia, primary, 32 OMIM
RSPH4A	21558	NM_001010892.2	99%	Ciliary dyskinesia, primary, 11 OMIM
RSPH9	21057	NM_152732.4	100%	Ciliary dyskinesia, primary, 12 OMIM
SBDS	19440	NM_016038.2	100%	Shwachman-Diamond syndrome OMIM
SCNN1A	10599	NM_001038.5	100%	Bronchiectasis with or without elevated sweat chloride 2 OMIM
SCNN1B	10600	NM_000336.2	100%	Bronchiectasis with or without elevated sweat chloride 1 OMIM
SCNN1G	10602	NM_001039.3	100%	Bronchiectasis with or without elevated sweat chloride 3 OMIM
SDCCAG8	10671	NM_006642.3	100%	Senior-Loken syndrome 7 OMIM Bardet-Biedl syndrome 16 OMIM
SPAG1	11212	NM_172218.2	99%	Ciliary dyskinesia, primary, 28 OMIM
SPATA7	20423	NM_018418.4	99%	Retinitis pigmentosa, juvenile, autosomal recessive OMIM Leber congenital amaurosis 3 OMIM
TCTN1	26113	NM_001082538.2	99%	Joubert syndrome 13 OMIM
TCTN2	25774	NM_024809.4	100%	Joubert syndrome 24 OMIM ?Meckel syndrome 8 OMIM
TCTN3	24519	NM_015631.5	100%	Orofaciodigital syndrome IV OMIM Joubert syndrome 18 OMIM
TMEM138	26944	NM_016464.4	100%	Joubert syndrome 16 OMIM
TMEM216	25018	NM_001173990.2	100%	Meckel syndrome 2 OMIM Joubert syndrome 2 OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
TMEM231	37234	NM_001077416.2	99%	Meckel syndrome 11 OMIM Joubert syndrome 20 OMIM
TMEM237	14432	NM_001044385.2	100%	Joubert syndrome 14 OMIM
TMEM67	28396	NM_153704.5	99%	Joubert syndrome 6 OMIM COACH syndrome OMIM {Bardet-Biedl syndrome 14, modifier of} OMIM Nephronophthisis 11 OMIM Meckel syndrome 3 OMIM
TOPORS	21653	NM_005802.4	100%	Retinitis pigmentosa 31 OMIM
TRIM32	16380	NM_012210.3	100%	?Bardet-Biedl syndrome 11 OMIM
TTC21B	25660	NM_024753.4	100%	Short-rib thoracic dysplasia 4 with or without polydactyly OMIM Nephronophthisis 12 OMIM
TTC8	20087	NM_198309.3	100%	Bardet-Biedl syndrome 8 OMIM ?Retinitis pigmentosa 51 OMIM
TULP1	12423	NM_003322.4	99%	Leber congenital amaurosis 15 OMIM Retinitis pigmentosa 14 OMIM
UMOD	12559	NM_003361.3	100%	Glomerulocystic kidney disease with hyperuricemia and isosthenuria OMIM Medullary cystic kidney disease 2 OMIM
USH1C	12597	NM_005709.3	100%	Usher syndrome, type 1C OMIM
USH1G	16356	NM_173477.4	99%	Usher syndrome, type 1G OMIM
USH2A	12601	NM_206933.2	100%	Retinitis pigmentosa 39 OMIM Usher syndrome, type 2A OMIM
WDPCP	28027	NM_015910.5	100%	?Congenital heart defects, hamartomas of tongue, and polysyndactyly OMIM
WDR19	18340	NM_025132.3	100%	Nephronophthisis 13 OMIM ?Short-rib thoracic dysplasia 5 with or without polydactyly OMIM ?Cranioectodermal dysplasia 4 OMIM Senior-Loken syndrome 8 OMIM
WDR34	28296	NM_052844.3	100%	Short-rib thoracic dysplasia 11 with or without polydactyly OMIM
WDR35	29250	NM_001006657.1	99%	Cranioectodermal dysplasia 2 OMIM Short-rib thoracic dysplasia 7 with or without polydactyly OMIM

Gen (symbol)	Gen (HGNC ID)	Transkript	>10x	Fenotype
WDR60	21862	NM_018051.4	100%	Short-rib thoracic dysplasia 8 with or without polydactyly OMIM
XPNPEP3	28052	NM_022098.3	100%	Nephronophthisis-like nephropathy 1 OMIM
ZIC3	12874	NM_003413.3	100%	VACTERL association, X-linked OMIM Heterotaxy, visceral, 1, X-linked OMIM Congenital heart defects, nonsyndromic, 1, X-linked OMIM
ZMYND10	19412	NM_015896.2	100%	Ciliary dyskinesia, primary, 22 OMIM
ZNF423	16762	NM_015069.3	100%	Nephronophthisis 14 OMIM Joubert syndrome 19 OMIM