Oslo universitetssykehus
Avdeling for medisinsk genetikk

Ciliopati

Genpanel, versjon v06

Sangersekvensering for påvisning av den sykdomsgivende varianten i CEP290 c.2991+1655A>G er utført

* 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: **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 affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ACVR2B	174	NM_001106.3		1-11	Heterotaxy, visceral, 4, autosomal OMIM
AHI1	21575	NM_017651.4		3-28	Joubert syndrome-3 OMIM
AIPL1	359	NM_014336.4		1-6	Retinitis pigmentosa, juvenile OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM
ANKS6	26724	NM_173551.3		1-15	Nephronophthisis 16 OMIM
ARL13B	25419	NM_182896.2		1-10	Joubert syndrome 8 OMIM

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ARL6	13210	NM_177976.2		3-9	{Bardet-Biedl syndrome 1, modifier of} OMIM Bardet-Biedl syndrome 3 OMIM
ARMC4	25583	NM_018076.3	2-10	2-20	Ciliary dyskinesia, primary, 23 OMIM
ATXN10	10549	NM_013236.3		1-12	Spinocerebellar ataxia 10 OMIM
B9D1	24123	NM_015681.3		1-7	Joubert syndrome 27 OMIM
B9D2	28636	NM_030578.3		2-4	Meckel syndrome 10 OMIM
BBIP1	28093	NM_001195306.1		2-4	Bardet-Biedl syndrome 18 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	Retinitis pigmentosa 74 OMIM Bardet-Biedl syndrome 2 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
BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
C21orf59	1301	NM_021254.2		1-7	Ciliary dyskinesia, primary, 26 OMIM
C2CD3	24564	NM_015531.5		1-31	Orofaciodigital syndrome XIV OMIM

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C2orf71	34383	NM_001029883.2		1-2	Retinitis pigmentosa 54 OMIM
C5orf42	25801	NM_023073.3		2-52	Orofaciodigital syndrome VI OMIM Joubert syndrome 17 OMIM
CABP4	1386	NM_145200.3		1-6	Cone-rod synaptic disorder, congenital nonprogressive OMIM
CC2D2A	29253	NM_001080522.2		3-38	Meckel syndrome 6 OMIM Joubert syndrome 9 OMIM COACH syndrome OMIM
CCDC103	32700	NM_213607.2		2-4	Ciliary dyskinesia, primary, 17 OMIM
CCDC11	26530	NM_145020.3		1-8	Heterotaxy, visceral, 6, autosomal recessive OMIM
CCDC114	26560	NM_144577.3		2-14	Ciliary dyskinesia, primary, 20 OMIM
CCDC151	28303	NM_145045.4		1-13	Ciliary dyskinesia, primary, 30 OMIM
CCDC28B	28163	NM_024296.4		2-6	{Bardet-Biedl syndrome 1, modifier of} OMIM
CCDC39	25244	NM_181426.1		1-20	Ciliary dyskinesia, primary, 14 OMIM
CCDC40	26090	NM_017950.3		1-20	Ciliary dyskinesia, primary, 15 OMIM
CCDC41	17966	NM_016122.2		3-17	Nephronophthisis 18 OMIM
CCDC65	29937	NM_033124.4		1-8	Ciliary dyskinesia, primary, 27 OMIM
CCNO	18576	NM_021147.4		1-3	Ciliary dyskinesia, primary, 29 OMIM
CDH23	13733	NM_022124.5		2-68	Usher syndrome, type 1D/F digenic OMIM Usher syndrome, type 1D OMIM
CEP104	24866	NM_014704.3		2-22	Joubert syndrome 25 OMIM

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CEP120	26690	NM_153223.3		2-21	Short-rib thoracic dysplasia 13 with or without polydactyly OMIM
CEP164	29182	NM_014956.4		3-33	Nephronophthisis 15 OMIM
CEP290	29021	NM_025114.3	54	2-54	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		1-11	Joubert syndrome 15 OMIM
CLRN1	12605	NM_174878.2		1-3	Usher syndrome, type 3A OMIM Retinitis pigmentosa 61 OMIM
CRB1	2343	NM_201253.2		1-12	Retinitis pigmentosa-12, autosomal recessive OMIM Pigmented paravenous chorioretinal atrophy OMIM Leber congenital amaurosis 8 OMIM
CRELD1	14630	NM_015513.4		1-10	Atrioventricular septal defect, partial, with heterotaxy syndrome OMIM
CRX	2383	NM_000554.4		2-4	Cone-rod retinal dystrophy-2 OMIM Leber congenital amaurosis 7 OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
DCDC2	18141	NM_016356.4		1-10	Nephronophthisis 19 OMIM ?Deafness, autosomal recessive 66 OMIM
DDX59	25360	NM_001031725.4		2-8	Orofaciodigital syndrome V OMIM
DNAAF1	30539	NM_178452.4		1-12	Ciliary dyskinesia, primary, 13 OMIM
DNAAF2	20188	NM_018139.2		1-3	Ciliary dyskinesia, primary, 10 OMIM
DNAAF3	30492	NM_001256714.1		1-12	Ciliary dyskinesia, primary, 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
DNAH11	2942	NM_001277115.1	76, 82	1-82	Ciliary dyskinesia, primary, 7, with or without situs inversus OMIM
DNAH5	2950	NM_001369.2		1-79	Ciliary dyskinesia, primary, 3, with or without situs inversus OMIM
DNAI1	2954	NM_012144.3		1-20	Ciliary dyskinesia, primary, 1, with or without situs inversus OMIM
DNAI2	18744	NM_023036.4		2-13	Ciliary dyskinesia, primary, 9, with or without situs inversus OMIM
DNAJB13	30718	NM_153614.3		1-8	Ciliary dyskinesia, primary, 34 OMIM
DNAL1	23247	NM_031427.3		1-8	Ciliary dyskinesia, primary, 16 OMIM
DPH1	3003	NM_001383.3		1-12	Developmental delay with short stature, dysmorphic features, and sparse hair OMIM
DRC1	24245	NM_145038.3		1-17	Ciliary dyskinesia, primary, 21 OMIM
DYNC2H1	2962	NM_001080463.1		1-90	Short-rib thoracic dysplasia 3 with or without polydactyly OMIM
DYNC2LI1	24595	NM_001193464.1		1-13	Short-rib throacic dysplasia 15 with polydactyly OMIM
DYX1C1	21493	NM_130810.3		2-10	Ciliary dyskinesia, primary, 25 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
GAS8	4166	NM_001286209.1		2-11	Ciliary dyskinesia, primary, 33 OMIM
GDF6	4221	NM_001001557.2		1-2	Leber congenital amaurosis 17 OMIM Klippel-Feil syndrome 1, autosomal dominant OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GLI3	4319	NM_000168.5		2-15	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		1-6	Nephronophthisis 7 OMIM
GPR98	17416	NM_032119.3		1-90	Usher syndrome, type 2C OMIM Usher syndrome, type 2C, GPR98/PDZD7 digenic OMIM
GUCY2D	4689	NM_000180.3		2-19	Leber congenital amaurosis 1 OMIM Cone-rod dystrophy 6 OMIM
HEATR2	26013	NM_017802.3		1-13	Ciliary dyskinesia, primary, 18 OMIM
HNF1B	11630	NM_000458.3		1-9	Renal cysts and diabetes syndrome OMIM
HYLS1	26558	NM_145014.2		4	Hydrolethalus syndrome OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM
IFT140	29077	NM_014714.3		3-31	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT172	30391	NM_015662.2		1-48	Short-rib thoracic dysplasia 10 with or without polydactyly OMIM Retinitis pigmentosa 71 OMIM
IFT27	18626	NM_006860.4		1-7	?Bardet-Biedl syndrome 19 OMIM
IFT43	29669	NM_052873.2		1-8	Cranioectodermal dysplasia 3 OMIM
IFT52	15901	NM_016004.3		2-14	Short-rib thoracic dysplasia 16 with or without polydactyly OMIM
IFT74	21424	NM_001099223.1		2-20	?Bardet-Biedl syndrome 20 OMIM

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IFT80	29262	NM_020800.2		2-20	Short-rib thoracic dysplasia 2 with or without polydactyly OMIM
IMPDH1	6052	NM_000883.3		1-17	Retinitis pigmentosa 10 OMIM Leber congenital amaurosis 11 OMIM
INPP5E	21474	NM_019892.4		1-10	Mental retardation, truncal obesity, retinal dystrophy, and micropenis OMIM Joubert syndrome 1 OMIM
INVS	17870	NM_014425.3		2-17	Nephronophthisis 2, infantile OMIM
IQCB1	28949	NM_001023570.2		3-15	Senior-Loken syndrome 5 OMIM
KCNJ13	6259	NM_002242.4		2-3	Leber congenital amaurosis 16 OMIM
KIAA0556	29068	NM_015202.2		1-28	Joubert syndrome 26 OMIM
KIAA0586	19960	NM_001244189.1		1-34	Short-rib thoracic dysplasia 14 with polydactyly OMIM Joubert syndrome 23 OMIM
KIF7	30497	NM_198525.2		2-19	Joubert syndrome 12 OMIM Acrocallosal syndrome OMIM
LBR	6518	NM_002296.3		2-14	Pelger-Huet anomaly OMIM
LCA5	31923	NM_181714.3		3-9	Leber congenital amaurosis 5 OMIM
LRAT	6685	NM_004744.4		2-3	Retinitis pigmentosa, juvenile OMIM Retinal dystrophy, early-onset severe OMIM Leber congenital amaurosis 14 OMIM
LRRC6	16725	NM_012472.4		1-12	Ciliary dyskinesia, primary, 19 OMIM
LZTFL1	6741	NM_020347.3		1-10	Bardet-Biedl syndrome 17 OMIM
MAPKBP1	29536	NM_014994.2		2-31	Nephronophthisis 20 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MCIDAS	40050	NM_001190787.1		1-7	Mucociliary clearance disorder
MKKS	7108	NM_018848.3		3-6	McKusick-Kaufman syndrome OMIM Bardet-Biedl syndrome 6 OMIM
MKS1	7121	NM_017777.3		1-18	Bardet-Biedl syndrome 13 OMIM Meckel syndrome 1 OMIM Joubert syndrome 28 OMIM
MUC1	7508	NM_002456.5		1-8	Medullary cystic kidney disease 1 OMIM
MYO7A	7606	NM_000260.3		2-49	Usher syndrome, type 1B OMIM
NEK1	7744	NM_012224.2		2-34	Short-rib thoracic dysplasia 6 with or without polydactyly OMIM
NEK8	13387	NM_178170.2		1-15	?Renal-hepatic-pancreatic dysplasia 2 OMIM ?Nephronophthisis 9 OMIM
NME8	16473	NM_016616.4		3-17	Ciliary dyskinesia, primary, 6 OMIM
NMNAT1	17877	NM_022787.3		2-5	Leber congenital amaurosis 9 OMIM
NODAL	7865	NM_018055.4		1-3	Heterotaxy, visceral, 5 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
OFD1	2567	NM_003611.2		1-23	Orofaciodigital syndrome I OMIM Joubert syndrome 10 OMIM ?Retinitis pigmentosa 23 OMIM Simpson-Golabi-Behmel syndrome, type 2 OMIM

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PCDH15	14674	NM_033056.3		2-33	Usher syndrome, type 1F OMIM Usher syndrome, type 1D/F digenic OMIM
PDE6D	8788	NM_002601.3		1-5	?Joubert syndrome 22 OMIM
PKHD1	9016	NM_138694.3		2-67	Polycystic kidney and hepatic disease OMIM
PRPH2	9942	NM_000322.4		1-3	Leber congenital amaurosis 18 OMIM Retinitis pigmentosa 7 and digenic OMIM
RD3	19689	NM_183059.2		2-3	Leber congenital amaurosis 12 OMIM
RDH12	19977	NM_152443.2		3-9	Leber congenital amaurosis 13 OMIM
REN	9958	NM_000537.3		1-10	Renal tubular dysgenesis OMIM
RPE65	10294	NM_000329.2		1-14	Retinitis pigmentosa 20 OMIM Leber congenital amaurosis 2 OMIM
RPGR	10295	NM_001034853.1		1-15	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness OMIM Retinitis pigmentosa 3 OMIM
RPGRIP1	13436	NM_020366.3		1-24	Leber congenital amaurosis 6 OMIM
RPGRIP1L	29168	NM_015272.2		2-27	Meckel syndrome 5 OMIM Joubert syndrome 7 OMIM COACH syndrome OMIM
RSPH1	12371	NM_080860.3		1-9	Ciliary dyskinesia, primary, 24 OMIM
RSPH3	21054	NM_031924.4		1-8	Ciliary dyskinesia, primary, 32 OMIM
RSPH4A	21558	NM_001010892.2		1-6	Ciliary dyskinesia, primary, 11 OMIM
RSPH9	21057	NM_152732.4		1-5	Ciliary dyskinesia, primary, 12 OMIM

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SBDS	19440	NM_016038.2	1-5	1-5	Shwachman-Diamond syndrome OMIM
SCNN1A	10599	NM_001038.5		2-13	Bronchiectasis with or without elevated sweat chloride 2 OMIM
SCNN1B	10600	NM_000336.2		2-13	Bronchiectasis with or without elevated sweat chloride 1 OMIM
SCNN1G	10602	NM_001039.3		2-13	Bronchiectasis with or without elevated sweat chloride 3 OMIM
SDCCAG8	10671	NM_006642.3		1-18	Senior-Loken syndrome 7 OMIM Bardet-Biedl syndrome 16 OMIM
SPAG1	11212	NM_172218.2		2-19	Ciliary dyskinesia, primary, 28 OMIM
SPATA7	20423	NM_018418.4		1-12	Retinitis pigmentosa, juvenile, autosomal recessive OMIM Leber congenital amaurosis 3 OMIM
TCTN1	26113	NM_001082538.2		1-14	Joubert syndrome 13 OMIM
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
TMEM138	26944	NM_016464.4		2-5	Joubert syndrome 16 OMIM
TMEM216	25018	NM_001173990.2		1-5	Meckel syndrome 2 OMIM Joubert 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 {Bardet-Biedl syndrome 14, modifier of} OMIM Nephronophthisis 11 OMIM Meckel syndrome 3 OMIM

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TOPORS	21653	NM_005802.4		1-3	Retinitis pigmentosa 31 OMIM
TRIM32	16380	NM_012210.3		2	?Bardet-Biedl syndrome 11 OMIM
TTC21B	25660	NM_024753.4		1-29	Short-rib thoracic dysplasia 4 with or without polydactyly OMIM Nephronophthisis 12 OMIM
TTC8	20087	NM_198309.3		2-15	Bardet-Biedl syndrome 8 OMIM ?Retinitis pigmentosa 51 OMIM
TULP1	12423	NM_003322.4		1-15	Leber congenital amaurosis 15 OMIM Retinitis pigmentosa 14 OMIM
UMOD	12559	NM_003361.3		2-11	Glomerulocystic kidney disease with hyperuricemia and isosthenuria OMIM Medullary cystic kidney disease 2 OMIM
USH1C	12597	NM_005709.3		1-21	Usher syndrome, type 1C OMIM
USH1G	16356	NM_173477.4		1-3	Usher syndrome, type 1G OMIM
USH2A	12601	NM_206933.2		2-72	Retinitis pigmentosa 39 OMIM Usher syndrome, type 2A OMIM
WDPCP	28027	NM_015910.5		1-18	?Congenital heart defects, hamartomas of tongue, and polysyndactyly OMIM
WDR19	18340	NM_025132.3		1-36	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		1-9	Short-rib thoracic dysplasia 11 with or without polydactyly OMIM
WDR35	29250	NM_001006657.1		1-28	Cranioectodermal dysplasia 2 OMIM Short-rib thoracic dysplasia 7 with or without polydactyly OMIM

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WDR60	21862	NM_018051.4		1-25	Short-rib thoracic dysplasia 8 with or without polydactyly OMIM
XPNPEP3	28052	NM_022098.3		1-10	Nephronophthisis-like nephropathy 1 OMIM
ZIC3	12874	NM_003413.3		1-3	VACTERL association, X-linked OMIM Heterotaxy, visceral, 1, X-linked OMIM Congenital heart defects, nonsyndromic, 1, X-linked OMIM
ZMYND10	19412	NM_015896.2		1-12	Ciliary dyskinesia, primary, 22 OMIM
ZNF423	16762	NM_015069.3		1-8	Nephronophthisis 14 OMIM Joubert syndrome 19 OMIM