Whole Exome Sequencing
Gene package Ciliopathy, version 5, 30-9-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751	602730	105	100	100	98
AHI1	Joubert syndrome 3, 608629	608894	67	100	98	90
ALMS1	Alstrom syndrome, 203800	606844	100	100	100	99
ANKS6	Nephronophthisis 16, 615382	615370	89	95	93	90
ARL13B	Joubert syndrome 8, 612291	608922	63	100	100	94
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	43	96	95	83
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	67	98	96	92
ARMC9	Joubert syndrome 30, 617622	617612	76	100	98	93
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	90	100	100	100
B9D2	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175	611951	82	100	100	100
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	45	100	96	86
BBS1	Bardet-Biedl syndrome 1, 209900	209901	126	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	66	100	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	59	100	100	99
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	77	100	100	97
BBS4	Bardet-Biedl syndrome 4, 615982	600374	76	100	100	95
BBS5	Bardet-Biedl syndrome 5, 615983	603650	65	100	98	89

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BBS7	Bardet-Biedl syndrome 7, 615984	607590	64	100	99	95
BBS9	Bardet-Biedl syndrome 9, 615986	607968	59	96	95	90
C2CD3	Orofaciodigital syndrome XIV, 615948	615944	80	100	100	99
C8orf37	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500	614477	78	100	100	96
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	66	100	100	95
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	83	100	100	100
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	125	100	100	100
CCDC151	Ciliary dyskinesia, primary, 30, 616037	615956	104	100	100	100
CCDC28B	{Bardet-Biedl syndrome 1, modifier of}, 209900	610162	83	100	100	100
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	80	100	99	94
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	100	100	100	100
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	57	100	100	94
CCNO	Ciliary dyskinesia, primary, 29, 615872	607752	113	100	100	100
CENPF	Stromme syndrome, 243605	600236	70	100	99	95
CEP104	Joubert syndrome 25, 616781	616690	71	100	98	93
CEP120	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	75	100	100	96
CEP164	Nephronophthisis 15, 614845	614848	93	100	100	98
CEP290	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189	610142	71	100	98	89
CEP41	Joubert syndrome 15, 614464	610523	69	100	100	94
CEP55	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500	610000	71	100	100	97
CEP83	Nephronophthisis 18, 615862	615847	60	100	96	81
CFAP300	Ciliary dyskinesia, primary, 38, 618063	0	53	100	98	87
CFAP410	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271	0	141	100	100	100
CFC1	Heterotaxy, visceral, 2, autosomal, 605376	618058	70	41	34	28
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	0	71	100	99	95
CSPP1	Joubert syndrome 21, 615636	603191	83	100	100	97
DCDC2	?Deafness 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394	605194	96	100	100	97

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DDX59	Orofaciodigital syndrome V, 174300	614571	62	100	98	92
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	611654	131	100	100	99
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	605755	124	100	100	100
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	615464	98	100	100	99
DNAAF4	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700	613190	54	100	99	89
DNAAF5	Ciliary dyskinesia, primary, 18, 614874	612517	104	96	86	79
DNAH1	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576	614566	127	100	100	100
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	608706	74	100	100	96
DNAH17	No OMIM phenotype	614864	94	100	99	96
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603332	74	100	100	96
DNAH9	Ciliary dyskinesia, primary, 40, 618300	603339	93	100	100	98
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	610063	91	100	98	97
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	603335	131	100	100	99
DNAJB13	Ciliary dyskinesia, primary, 34, 617091	603330	88	100	100	100
DNAL1	Ciliary dyskinesia, primary, 16, 614017	604366	77	100	100	95
DNHD1	No OMIM phenotype	605483	117	100	100	100
DRC1	Ciliary dyskinesia, primary, 21, 615294	610263	81	100	100	98
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	610062	71	100	99	91
DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly, 617088	617277	53	100	100	91
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	615288	98	96	94	94
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	603297	87	100	99	95
EXOC8	No OMIM phenotype	617083	118	100	100	100
FAM149B1	No OMIM phenotype	604831	61	100	93	83
FUZ	{Neural tube defects, susceptibility to}, 182940	607261	109	100	100	100
GAS2L2	?Ciliary dyskinesia, primary, 41, 618449	615283	154	100	100	100
GAS8	Ciliary dyskinesia, primary, 33, 616726	618413	85	99	97	93
GDF1	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530	610622	69	100	93	81
GLIS2	Nephronophthisis 7, 611498	611398	117	100	100	100
HYDIN	Ciliary dyskinesia, primary, 5, 608647	605178	46	82	69	57
HYLS1	Hydrolethalus syndrome, 236680	602880	74	100	100	100
IFT122	Cranioectodermal dysplasia 1, 218330	608539	111	100	100	99
IFT140	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	610812	113	100	100	98
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	610693	75	100	100	96

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IFT27	?Bardet-Biedl syndrome 19, 615996	606045	84	100	100	99
IFT43	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866	614620	75	100	100	100
IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102	607386	50	100	99	91
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	615870	62	100	98	87
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895	614068	47	99	94	82
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	617094	113	100	100	97
INTU	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925	611177	59	100	99	92
INVS	Nephronophthisis 2, infantile, 602088	605489	81	100	100	97
IQCB1	Senior-Loken syndrome 5, 609254	613037	54	100	94	78
KCTD3	No OMIM phenotype	610621	61	100	100	96
KIAA0556	Joubert syndrome 26, 616784	243305	97	100	99	97
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	609237	72	100	98	94
KIAA0753	?Orofaciodigital syndrome XV, 617127	613272	52	100	98	91
KIF14	?Meckel syndrome 12, 616258 Microcephaly 20, primary, 617914	616650	74	100	99	91
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	610178	107	98	96	93
LCA5	Leber congenital amaurosis 5, 604537	617112	74	100	100	99
LEFTY2	No OMIM phenotype	611279	144	100	100	99
LRRC56	Ciliary dyskinesia, primary, 19, 614935	611254	118	100	100	98
LRRC6	Ciliary dyskinesia, primary, 39, 618254	611408	79	100	99	93
LZTFL1	Bardet-Biedl syndrome 17, 615994	601877	79	100	100	93
MAPKBP1	Nephronophthisis 20, 617271	614930	108	100	100	98
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	618227	80	100	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	606568	117	100	100	99
NCAPG2	Khan-Khan-Katsanis syndrome, 618460	616786	51	100	94	79
NEK1	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604896	56	100	98	89
NEK8	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415	609883	132	100	100	100

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NME8	Ciliary dyskinesia, primary, 6, 610852	608532	65	100	100	93
NODAL	Heterotaxy, visceral, 5, 270100	604588	119	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	609799	58	100	98	88
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	607421	66	100	99	94
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	601265	110	100	100	100
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	607100	43	100	95	80
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	608002	43	100	95	75
PDE6D	?Joubert syndrome 22, 615665	607215	86	100	100	100
PIBF1	Joubert syndrome 33, 617767	300535	42	100	93	72
PIH1D3	Ciliary dyskinesia, primary, 36, 300991	300170	40	100	92	66
PKD1	Polycystic kidney disease 1, 173900	602676	93	97	95	90
PKD2	Polycystic kidney disease 2, 613095	607532	62	100	97	89
PKHD1	Polycystic kidney disease 4, with or without hepatic disease, 263200	300933	82	100	100	98
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	601313	90	100	100	100
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	173910	88	100	100	96
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	606702	60	98	96	89
RSPH1	Ciliary dyskinesia, primary, 24, 615481	614783	80	100	100	95
RSPH3	Ciliary dyskinesia, primary, 32, 616481	605446	89	100	99	94
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	610937	100	100	100	100
RSPH9	Ciliary dyskinesia, primary, 12, 612650	609314	125	100	100	100
SCLT1	No OMIM phenotype	615876	51	100	97	87
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	612647	92	100	100	96
SPAG1	Ciliary dyskinesia, primary, 28, 615505	612648	64	100	99	91
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	611399	62	100	99	94

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SUFU	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174	613524	104	100	100	100
TBC1D32	No OMIM phenotype	603395	61	100	98	90
TCTN1	Joubert syndrome 13, 614173	609868	94	100	100	98
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	607035	84	100	100	97
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	615867	66	100	100	97
TMEM107	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563	609863	100	100	100	94
TMEM138	Joubert syndrome 16, 614465	613846	54	100	100	100
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613847	87	100	100	95
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	616183	96	100	95	90
TMEM237	Joubert syndrome 14, 614424	614459	57	100	98	87
TMEM260	Structural heart defects and renal anomalies syndrome, 617478	613277	58	100	98	93
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 ?RHYNS syndrome, 602152	614949	77	100	100	94
TRAF3IP1	Senior-Loken syndrome 9, 616629	614423	55	100	97	86
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle 8, 254110	617449	107	100	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	609884	74	100	100	99
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	607380	75	100	99	94
TTC25	Ciliary dyskinesia, primary, 35, 617092	602290	71	100	99	97
TTC26	No OMIM phenotype	611695	55	100	99	93
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	612014	66	100	100	93
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	617095	108	100	100	100

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VHL	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300	617453	183	100	100	100
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	608132	72	100	99	93
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	602280	71	100	100	97
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	608537	116	100	100	100
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613580	62	100	100	94
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	608151	80	100	99	94
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613363	63	100	100	96
ZIC3	Congenital heart defects, nonsyndromic, 1, 306955 Heterotaxy, visceral, 1, 306955 VACTERL association, 314390	613602	101	100	100	97
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	615462	130	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	613553	166	100	100	100

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x