Whole Exome Sequencing
Gene package Ciliopathy, version 2, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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 sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751	602730	93	100	96
AHI1	Joubert syndrome 3, 608629	608894	77	100	98
ALMS1	Alstrom syndrome, 203800	606844	105	100	100
ANKS6	Nephronophthisis 16, 615382	615370	63	93	89
ARL13B	Joubert syndrome 8, 612291	608922	61	100	100
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	48	95	81
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	40	93	82
ATXN10	Spinocerebellar ataxia 10, 603516	611150	65	100	97
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	84	100	100
B9D2	Meckel syndrome 10, 614175	611951	74	100	100
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	53	100	89
BBS1	Bardet-Biedl syndrome 1, 209900	209901	92	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	76	100	100
BBS12	Bardet-Biedl syndrome 12, 615989	610683	98	100	100
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	76	100	98
BBS4	Bardet-Biedl syndrome 4, 615982	600374	80	100	99
BBS5	Bardet-Biedl syndrome 5, 615983	603650	59	99	92
BBS7	Bardet-Biedl syndrome 7, 615984	607590	68	100	98

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BBS9	Bardet-Biedl syndrome 9, 615986	607968	76	98	93
C21orf59	Ciliary dyskinesia, primary, 26, 615500	615494	44	92	85
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	47	98	91
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	78	100	96
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	82	100	99
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	95	100	100
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	59	100	94
CCDC151	Ciliary dyskinesia, primary, 30, 616037	615956	55	100	94
CCDC28B	{Bardet-Biedl syndrome 1, modifier of}, 209900	610162	59	100	100
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	62	100	96
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	83	100	100
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	29	94	71
CCNO	Ciliary dyskinesia, primary, 29, 615872	607752	48	99	92
CENPF	Stromme syndrome, 243605	600236	50	96	87
CEP104	Joubert syndrome 25, 616781	616690	47	99	88
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	47	98	87
CEP164	Nephronophthisis 15, 614845	614848	68	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	55	98	92
CEP41	Joubert syndrome 15, 614464	610523	64	100	93
CEP83	Nephronophthisis 18, 615862	615847	34	86	55
CFC1	Heterotaxy, visceral, 2, autosomal, 605376	605194	35	76	55
CSPP1	Joubert syndrome 21, 615636	611654	38	96	81
DCDC2	?Deafness 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394	605755	58	100	93
DDX59	Orofaciodigital syndrome V, 174300	615464	59	99	89
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	613190	88	100	100
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	612517	70	100	97
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	614566	70	100	97
DNAAF4	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700	608706	63	100	0
DNAAF5	Ciliary dyskinesia, primary, 18, 614874	614864	82	90	82
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	603339	87	100	99

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DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603335	85	100	99
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	604366	84	100	97
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	605483	107	100	98
DNAJB13	Ciliary dyskinesia, primary, 34, 617091	610263	42	99	79
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	57	90	89
DNHD1	No OMIM phenotype	617277	69	100	96
DRC1	Ciliary dyskinesia, primary, 21, 615294	615288	34	96	79
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	64	99	94
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	604831	77	94	91
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	78	97	95
EXOC8	No OMIM phenotype	615283	79	100	100
FLNC	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	102565	90	100	99
FOXH1	No OMIM phenotype	603621	50	100	100
GAS8	Ciliary dyskinesia, primary, 33, 616726	605178	48	99	90
GDF1	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854	602880	27	70	49
GLIS2	Nephronophthisis 7, 611498	608539	74	100	96
HYDIN	Ciliary dyskinesia, primary, 5, 608647	610812	147	100	100
HYLS1	Hydrolethalus syndrome, 236680	610693	66	100	100
IFT122	Cranioectodermal dysplasia 1, 218330	606045	97	100	99
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	82	100	99
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	77	100	99
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	45	100	83
IFT43	Cranioectodermal dysplasia 3, 614099	614068	45	100	99
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	54	99	92
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	71	98	92
INVS	Nephronophthisis 2, infantile, 602088	243305	82	100	99
IQCB1	Senior-Loken syndrome 5, 609254	609237	65	99	89
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	39	89	75
KIF14	?Meckel syndrome 12, 616258	611279	43	90	70

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KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	64	98	93
LCA5	Leber congenital amaurosis 5, 604537	611408	70	100	97
LEFTY2	Left-right axis malformations	601877	31	81	65
LRRC6	Ciliary dyskinesia, primary, 19, 614935	614930	79	100	95
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	87	100	92
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	106	100	100
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	73	100	98
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	62	99	92
NEK8	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415	609799	94	100	100
NME8	Ciliary dyskinesia, primary, 6, 610852	607421	76	100	99
NODAL	Heterotaxy, visceral, 5, 270100	601265	106	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	78	100	99
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	77	100	100
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	87	100	100
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	45	99	93
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	43	99	88
PDE6D	?Joubert syndrome 22, 615665	602676	43	100	94
PKD1	Polycystic kidney disease, adult type I, 173900	601313	76	98	95
PKD2	Polycystic kidney disease 2, 613095	173910	67	95	89
PKHD1	Polycystic kidney and hepatic disease, 263200	606702	83	100	99
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	90	100	100
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	86	100	99

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RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	77	97	95
RSPH1	Ciliary dyskinesia, primary, 24, 615481	609314	54	100	94
RSPH3	Ciliary dyskinesia, primary, 32, 616481	615876	50	97	86
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	612647	78	100	100
RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	100	100	100
SCLT1	No OMIM phenotype	611399	60	100	95
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	70	100	99
SPAG1	Ciliary dyskinesia, primary, 28, 615505	603395	36	89	69
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	71	100	96
TBC1D32	No OMIM phenotype	615867	30	88	60
TCTN1	Joubert syndrome 13, 614173	609863	70	100	97
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	93	100	100
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	60	100	96
TMEM138	Joubert syndrome 16, 614465	614459	67	100	100
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	87	100	100
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	56	98	93
TMEM237	Joubert syndrome 14, 614424	614423	80	100	99
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	51	99	90
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	77	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	611695	70	100	99
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	70	100	97
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	64	100	90
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	65	100	97

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VHL	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300	608537	75	98	87
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	67	100	93
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	73	100	98
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	48	100	93
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	82	100	95
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	41	94	81
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	85	100	99
ZIC3	Congenital heart defects, nonsyndromic, 1, 306955 Heterotaxy, visceral, 1, 306955 VACTERL association, 314390	300265	53	100	100
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	607070	60	100	96
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	102	100	100

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x