Whole Exome Sequencing
Gene package Ciliopathy, version 2.1, 22-11-2017

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	74	100	100	96
AHI1	Joubert syndrome 3, 608629	608894	51	100	94	73
ALMS1	Alstrom syndrome, 203800	606844	82	100	99	96
ANKS6	Nephronophthisis 16, 615382	615370	56	94	91	82
ARL13B	Joubert syndrome 8, 612291	608922	50	100	99	86
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	32	97	88	57
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	50	100	98	84
ATXN10	Spinocerebellar ataxia 10, 603516	611150	43	100	97	78
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	60	100	100	96
B9D2	Meckel syndrome 10, 614175	611951	55	100	100	99
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	36	100	92	65
BBS1	Bardet-Biedl syndrome 1, 209900	209901	69	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	55	100	100	98
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	96
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	58	100	99	87
BBS4	Bardet-Biedl syndrome 4, 615982	600374	53	100	96	81
BBS5	Bardet-Biedl syndrome 5, 615983	603650	51	100	99	82

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BBS7	Bardet-Biedl syndrome 7, 615984	607590	51	100	98	87
BBS9	Bardet-Biedl syndrome 9, 615986	607968	42	96	94	79
C21orf59	Ciliary dyskinesia, primary, 26, 615500	615494	44	100	96	81
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	58	100	99	92
C5orf42	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	54	100	98	88
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	48	100	97	81
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	71	100	100	98
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	79	100	100	100
CCDC151	Ciliary dyskinesia, primary, 30, 616037	615956	64	100	100	99
CCDC28B	{Bardet-Biedl syndrome 1, modifier of}, 209900	610162	59	100	100	100
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	67	100	97	88
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	65	100	100	97
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	40	100	98	78
CCNO	Ciliary dyskinesia, primary, 29, 615872	607752	86	100	100	100
CENPF	Stromme syndrome, 243605	600236	56	100	99	93
CEP104	Joubert syndrome 25, 616781	616690	45	100	93	77
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	54	100	99	86
CEP164	Nephronophthisis 15, 614845	614848	63	100	96	88
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	57	100	98	84
CEP41	Joubert syndrome 15, 614464	610523	53	100	99	89
CEP83	Nephronophthisis 18, 615862	615847	48	100	93	71
CFC1	Heterotaxy, visceral, 2, autosomal, 605376	605194	130	100	96	83
CSPP1	Joubert syndrome 21, 615636	611654	58	100	99	89
DCDC2	?Deafness 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394	605755	60	100	99	90
DDX59	Orofaciodigital syndrome V, 174300	615464	50	99	96	83
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	613190	79	100	99	91
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	612517	89	100	100	100
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	614566	65	100	100	99
DNAAF4	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700	608706	43	100	93	71

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DNAAF5	Ciliary dyskinesia, primary, 18, 614874	614864	64	92	84	77
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	603339	54	100	98	87
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603335	51	100	98	85
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	604366	58	100	98	92
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	605483	75	100	100	98
DNAJB13	Ciliary dyskinesia, primary, 34, 617091	610263	55	100	100	91
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	55	100	100	90
DNHD1	No OMIM phenotype	617277	81	100	100	99
DRC1	Ciliary dyskinesia, primary, 21, 615294	615288	58	100	99	87
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	53	100	97	82
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	604831	62	96	94	92
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	55	100	97	90
EXOC8	No OMIM phenotype	615283	84	100	100	100
FLNC	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	102565	90	100	100	100
FOXH1	No OMIM phenotype	603621	75	100	100	100
GAS8	Ciliary dyskinesia, primary, 33, 616726	605178	59	100	97	90
GDF1	Double-outlet right ventricle, 217095 Right atrial isomerism, 208530 Tetralogy of Fallot, 187500 Transposition of great arteries, dextro-looped 3, 613854	602880	40	100	90	65
GLIS2	Nephronophthisis 7, 611498	608539	61	100	100	93
HYDIN	Ciliary dyskinesia, primary, 5, 608647	610812	45	100	91	71
HYLS1	Hydrolethalus syndrome, 236680	610693	57	100	100	100
IFT122	Cranioectodermal dysplasia 1, 218330	606045	72	100	100	95
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	70	100	97	91
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	52	100	98	82
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	54	100	100	96
IFT43	Cranioectodermal dysplasia 3, 614099	614068	54	100	100	92
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	50	100	93	75
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	80	100	100	96
INVS	Nephronophthisis 2, infantile, 602088	243305	56	100	98	89
IQCB1	Senior-Loken syndrome 5, 609254	609237	42	100	89	67

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KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	54	100	96	86
KIF14	?Meckel syndrome 12, 616258	611279	59	100	96	82
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	66	99	95	89
LCA5	Leber congenital amaurosis 5, 604537	611408	63	100	100	97
LEFTY2	Left-right axis malformations	601877	87	100	100	100
LRRC6	Ciliary dyskinesia, primary, 19, 614935	614930	58	100	98	83
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	57	100	98	83
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	59	100	100	99
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	81	100	100	97
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	45	100	95	75
NEK8	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415	609799	92	100	100	100
NME8	Ciliary dyskinesia, primary, 6, 610852	607421	50	100	98	83
NODAL	Heterotaxy, visceral, 5, 270100	601265	79	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	39	100	92	67
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	47	100	97	82
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	67	100	100	98
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	36	100	92	67
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	42	100	97	81
PDE6D	?Joubert syndrome 22, 615665	602676	52	100	100	99
PKD1	Polycystic kidney disease, adult type I, 173900	601313	69	97	94	88
PKD2	Polycystic kidney disease 2, 613095	173910	45	100	96	72
PKHD1	Polycystic kidney and hepatic disease, 263200	606702	58	100	98	89

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POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	55	100	100	98
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	59	100	98	86
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	47	97	94	77
RSPH1	Ciliary dyskinesia, primary, 24, 615481	609314	45	100	96	79
RSPH3	Ciliary dyskinesia, primary, 32, 616481	615876	72	100	98	91
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	612647	73	100	100	96
RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	85	100	100	99
SCLT1	No OMIM phenotype	611399	40	100	95	72
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	66	100	99	87
SPAG1	Ciliary dyskinesia, primary, 28, 615505	603395	45	100	95	73
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, 604232	609868	51	100	98	89
TBC1D32	No OMIM phenotype	615867	45	100	96	76
TCTN1	Joubert syndrome 13, 614173	609863	61	100	99	88
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	60	100	98	90
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	50	100	99	90
TMEM138	Joubert syndrome 16, 614465	614459	38	100	100	87
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	61	100	99	76
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	71	100	100	90
TMEM237	Joubert syndrome 14, 614424	614423	38	100	90	69
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	58	100	98	82
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	86	100	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	611695	59	100	100	95
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	56	100	97	84

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TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	46	100	99	85
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	69	100	100	98
VHL	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300	608537	88	100	100	100
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	53	100	98	84
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	50	100	98	85
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	72	100	100	97
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	43	100	97	77
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	56	100	98	85
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	41	100	98	82
ZIC3	Congenital heart defects, nonsyndromic, 1, 306955 Heterotaxy, visceral, 1, 306955 VACTERL association, 314390	300265	60	100	97	86
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	607070	92	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	121	100	100	99

- 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 96 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x