Whole Exome Sequencing
Gene package Ciliopathy, version 4, 30-7-2018

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

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

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	100	100	100	97
AHI1	Joubert syndrome 3, 608629	608894	58	100	96	82
ALMS1	Alstrom syndrome, 203800	606844	84	100	99	97
ANKS6	Nephronophthisis 16, 615382	615370	84	95	93	88
ARL13B	Joubert syndrome 8, 612291	608922	53	100	99	90
ARL6	{Bardet-Biedl syndrome 1, modifier of}, 209900 Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575	608845	35	96	93	71
ARMC4	Ciliary dyskinesia, primary, 23, 615451	615408	62	100	99	90
ARMC9	Joubert syndrome 30, 617622	617612	68	100	97	88
ATXN10	Spinocerebellar ataxia 10, 603516	611150	52	100	99	86
B9D1	Joubert syndrome 27, 617120 ?Meckel syndrome 9, 614209	614144	83	100	100	100
B9D2	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175	611951	73	100	100	100
BBIP1	?Bardet-Biedl syndrome 18, 615995	613605	38	100	90	77
BBS1	Bardet-Biedl syndrome 1, 209900	209901	109	100	100	100
BBS10	Bardet-Biedl syndrome 10, 615987	610148	57	100	100	99
BBS12	Bardet-Biedl syndrome 12, 615989	610683	51	100	100	98
BBS2	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562	606151	68	100	100	93

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BBS4	Bardet-Biedl syndrome 4, 615982	600374	68	100	99	91
BBS5	Bardet-Biedl syndrome 5, 615983	603650	57	100	97	83
BBS7	Bardet-Biedl syndrome 7, 615984	607590	56	100	99	92
BBS9	Bardet-Biedl syndrome 9, 615986	607968	49	96	93	85
C2CD3	?Orofaciodigital syndrome XIV, 615948	615944	67	100	100	96
CC2D2A	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284	612013	56	100	99	90
CCDC103	Ciliary dyskinesia, primary, 17, 614679	614677	77	100	100	98
CCDC114	Ciliary dyskinesia, primary, 20, 615067	615038	120	100	100	100
CCDC151	Ciliary dyskinesia, primary, 30, 616037	615956	95	100	100	99
CCDC28B	{Bardet-Biedl syndrome 1, modifier of}, 209900	610162	75	100	100	97
CCDC39	Ciliary dyskinesia, primary, 14, 613807	613798	68	100	98	91
CCDC40	Ciliary dyskinesia, primary, 15, 613808	613799	90	100	100	99
CCDC65	Ciliary dyskinesia, primary, 27, 615504	611088	48	100	99	87
CCNO	Ciliary dyskinesia, primary, 29, 615872	607752	116	100	100	100
CENPF	Stromme syndrome, 243605	600236	60	100	99	90
CEP104	Joubert syndrome 25, 616781	616690	64	100	97	88
CEP120	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300	613446	63	100	99	92
CEP164	Nephronophthisis 15, 614845	614848	85	100	100	96
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	59	100	96	81
CEP41	Joubert syndrome 15, 614464	610523	61	100	99	92
CEP83	Nephronophthisis 18, 615862	615847	50	100	92	73
CFAP298	Ciliary dyskinesia, primary, 26, 615500	615494	63	100	99	93
CPLANE1	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170	614571	59	100	98	90
CSPP1	Joubert syndrome 21, 615636	611654	73	100	99	93
DCDC2	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394	605755	86	100	99	93
DDX59	Orofaciodigital syndrome V, 174300	615464	53	100	97	88
DNAAF1	Ciliary dyskinesia, primary, 13, 613193	613190	117	100	100	97
DNAAF2	Ciliary dyskinesia, primary, 10, 612518	612517	117	100	100	99
DNAAF3	Ciliary dyskinesia, primary, 2, 606763	614566	92	100	100	99

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DNAAF4	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700	608706	No coverage data			
DNAAF5	Ciliary dyskinesia, primary, 18, 614874	614864	99	97	89	79
DNAH1	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576	603332	116	100	100	100
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	603339	63	100	99	92
DNAH17	No OMIM phenotype	610063	87	100	99	95
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	603335	63	100	99	91
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	604366	82	100	98	96
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	605483	119	100	100	99
DNAJB13	Ciliary dyskinesia, primary, 34, 617091	610263	73	100	100	97
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	67	100	100	89
DNHD1	No OMIM phenotype	617277	105	100	100	99
DRC1	Ciliary dyskinesia, primary, 21, 615294	615288	71	100	100	96
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	603297	61	100	97	84
EVC	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530	604831	92	96	95	93
EVC2	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530	607261	80	100	99	94
EXOC8	No OMIM phenotype	615283	109	100	100	100
FLNC	Cardiomyopathy, familial hypertrophic, 26 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524	102565	124	100	100	99
FOXH1	No OMIM phenotype	603621	83	100	100	100
FUZ	Neural tube defects, 182940	610622	94	100	100	100
GAS8	Ciliary dyskinesia, primary, 33, 616726	605178	73	99	96	90
GDF1	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530	602880	68	100	93	81
GLIS2	Nephronophthisis 7, 611498	608539	106	100	100	100
HYDIN	Ciliary dyskinesia, primary, 5, 608647	610812	58	100	95	82
HYLS1	Hydrolethalus syndrome, 236680	610693	61	100	100	100
IFT122	Cranioectodermal dysplasia 1, 218330	606045	108	100	100	98
IFT140	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920	614620	110	100	99	97
IFT172	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	607386	65	100	99	92
IFT27	?Bardet-Biedl syndrome 19, 615996	615870	75	100	100	99

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IFT43	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866	614068	70	100	100	98
IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102	617094	42	100	98	82
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	611177	55	100	96	80
INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	115	100	100	98
INTU	?Orofaciodigital syndrome XVII, 617926 ?Short-rib throacic dysplasia 20 with polydactyly, 617925	610621	49	100	97	85
INVS	Nephronophthisis 2, infantile, 602088	243305	74	100	99	94
IQCB1	Senior-Loken syndrome 5, 609254	609237	48	100	92	74
KCTD3	No OMIM phenotype	613272	52	100	99	93
KIAA0556	Joubert syndrome 26, 616784	616650	86	100	99	96
KIAA0586	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546	610178	62	100	97	91
KIAA0753	?Orofaciodigital syndrome XV, 617127	617112	45	100	96	83
KIF14	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914	611279	63	100	97	87
KIF7	Acrocallosal syndrome, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Joubert syndrome 12, 200990	611254	104	99	96	93
LCA5	Leber congenital amaurosis 5, 604537	611408	63	100	100	97
LEFTY2	No OMIM phenotype	601877	138	100	100	100
LRRC6	Ciliary dyskinesia, primary, 19, 614935	614930	68	100	98	88
LZTFL1	Bardet-Biedl syndrome 17, 615994	606568	66	100	100	89
MAPKBP1	Nephronophthisis 20, 617271	616786	95	100	100	98
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	67	100	100	98
MKS1	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000	609883	112	100	100	98
NEK1	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520	604588	50	100	97	81
NEK8	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415	609799	122	100	100	100
NME8	Ciliary dyskinesia, primary, 6, 610852	607421	57	100	98	88
NODAL	Heterotaxy, visceral, 5, 270100	601265	116	100	100	100

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NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	50	100	96	82
NPHP3	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540	608002	57	100	98	88
NPHP4	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996	607215	105	100	100	99
OCRL	Dent disease 2, 300555 Lowe syndrome, 309000	300535	35	100	91	67
OFD1	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Simpson-Golabi-Behmel syndrome, type 2, 300209	300170	37	100	89	63
PDE6D	?Joubert syndrome 22, 615665	602676	74	100	100	100
PIBF1	Joubert syndrome 33, 617767	607532	38	100	91	65
PIH1D3	Ciliary dyskinesia, primary, 36, X-linked, 300991	300933	34	100	87	60
PKD1	Polycystic kidney disease 1, 173900	601313	101	97	96	94
PKD2	Polycystic kidney disease 2, 613095	173910	55	100	97	82
PKHD1	Polycystic kidney disease 4, with or without hepatic disease, 263200	606702	70	100	99	95
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	614783	78	100	100	100
RPGRIP1	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826	605446	76	100	99	93
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	52	98	94	81
RSPH1	Ciliary dyskinesia, primary, 24, 615481	609314	65	100	99	90
RSPH3	Ciliary dyskinesia, primary, 32, 616481	615876	85	100	98	93
RSPH4A	Ciliary dyskinesia, primary, 11, 612649	612647	90	100	100	99
RSPH9	Ciliary dyskinesia, primary, 12, 612650	612648	121	100	100	100
SCLT1	No OMIM phenotype	611399	44	100	95	77
SDCCAG8	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615	613524	78	100	99	90
SPAG1	Ciliary dyskinesia, primary, 28, 615505	603395	58	100	97	86
SPATA7	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232	609868	55	100	98	90

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SUFU	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174	607035	90	100	100	100
TBC1D32	No OMIM phenotype	615867	54	100	97	83
TCTN1	Joubert syndrome 13, 614173	609863	87	100	100	94
TCTN2	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885	613846	73	100	100	94
TCTN3	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860	613847	59	100	100	94
TMEM107	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563	616183	83	100	99	91
TMEM138	Joubert syndrome 16, 614465	614459	46	100	100	95
TMEM216	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194	613277	89	100	100	89
TMEM231	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397	614949	101	100	100	99
TMEM237	Joubert syndrome 14, 614424	614423	48	100	96	83
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	68	100	99	87
TRAF3IP1	Senior-Loken syndrome 9, 616629	607380	48	100	94	77
TRIM32	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110	602290	96	100	100	100
TTBK2	Spinocerebellar ataxia 11, 604432	611695	63	100	100	97
TTC21B	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819	612014	63	100	98	89
TTC26	No OMIM phenotype	617453	45	100	97	85
TTC8	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464	608132	57	100	99	87
TULP1	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132	602280	103	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	608537	170	100	100	100
WDPCP	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	613580	65	100	98	88
WDR19	?Cranioectodermal dysplasia 4, 614378 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376	608151	62	100	99	92
WDR34	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	613363	109	100	100	100
WDR35	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	613602	52	100	98	86
WDR60	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	615462	67	100	99	91
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159	613553	56	100	99	92
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390	300265	87	100	100	95
ZMYND10	Ciliary dyskinesia, primary, 22, 615444	607070	118	100	100	100
ZNF423	Joubert syndrome 19, 614844 Nephronophthisis 14, 614844	604557	161	100	100	100

- Gene symbols according HGCN
- OMIM release used: 4-7-2018
- "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
- % Covered 30x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 30x