Whole Exome Sequencing
Gene package Ciliopathy, version 7, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique 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 Alissa Interpret 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 sequence changes).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ACVR2B	602730	272	100	100	100
ADAMTS9	605421	142	100	99	98
AHI1	608894	65	96	86	77
ALMS1	606844	116	100	99	96
ANKS6	615370	196	99	93	92
ARL13B	608922	56	100	90	72
ARL6	608845	44	98	88	79
ARMC4	615408	81	92	85	78
ARMC9	617612	134	96	93	90
B9D1	614144	285	100	100	100
B9D2	611951	336	100	100	100
BBIP1	613605	79	100	99	92
BBS1	209901	225	100	100	100
BBS10	610148	100	100	100	99
BBS12	610683	118	100	100	98
BBS2	606151	117	100	99	94
BBS4	600374	100	100	100	93
BBS5	603650	52	91	66	47
BBS7	607590	68	95	82	75
BBS9	607968	73	99	94	88
C2CD3	615944	132	100	100	99
C8orf37	614477	124	100	93	85
CC2D2A	612013	88	98	93	86
CCDC103	614677	311	100	100	100
CCDC114	615038	213	100	100	100

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CCDC151	615956	186	100	100	100
CCDC28B	610162	164	100	100	99
CCDC39	613798	44	80	58	47
CCDC40	613799	166	100	100	100
CCDC65	611088	89	100	98	93
CCNO	607752	259	100	100	100
CENPF	600236	78	100	96	89
CEP104	616690	94	100	99	95
CEP120	613446	84	99	94	89
CEP164	614848	149	100	100	100
CEP290	610142	35	74	58	45
CEP41	610523	83	100	94	91
CEP55	610000	44	97	78	62
CEP83	615847	35	83	64	46
CFAP298	615494	113	100	91	87
CFAP300	618058	57	83	61	57
CFAP410	603191	250	100	100	100
CFAP53	614759	78	89	80	74
CFC1	605194	59	33	33	30
CPLANE1	614571	75	97	93	86
CSPP1	611654	68	99	91	80
DCDC2	605755	144	95	84	77
DDX59	615464	94	100	98	93
DNAAF1	613190	146	100	100	97
DNAAF2	612517	213	100	99	97
DNAAF3	614566	243	100	100	100
DNAAF4	608706	55	82	72	64
DNAAF5	614864	217	100	97	96
DNAH1	603332	212	100	100	100
DNAH11	603339	89	100	96	89
DNAH17	610063	185	100	100	100
DNAH5	603335	101	99	97	93
DNAH9	603330	143	100	100	98
DNAI1	604366	151	100	99	97
DNAI2	605483	202	100	100	100
DNAJB13	610263	154	100	100	100
DNAL1	610062	80	90	82	71
DNHD1	617277	228	100	100	100
DRC1	615288	111	100	99	93
DYNC2H1	603297	57	94	84	73
DYNC2LI1	617083	71	97	83	77
EVC	604831	189	94	92	91
EVC2	607261	181	100	100	99

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EXOC8	615283	206	100	100	100
FAM149B1	618413	95	100	100	98
FOXJ1	602291	220	100	100	100
FUZ	610622	183	100	100	100
GAS2L2	611398	312	100	100	100
GAS8	605178	146	100	100	100
GDF1	602880	150	100	100	99
GLIS2	608539	245	100	100	100
HYDIN	610812	86	88	81	76
HYLS1	610693	114	100	100	100
IFT122	606045	141	100	100	98
IFT140	614620	199	100	100	100
IFT172	607386	143	100	100	98
IFT27	615870	101	100	100	99
IFT43	614068	133	100	100	100
IFT52	617094	72	100	99	89
IFT80	611177	49	92	82	69
IFT81	605489	37	96	78	54
INPP5E	613037	259	100	100	100
INTU	610621	87	100	95	88
INVS	243305	123	100	99	97
IQCB1	609237	75	100	99	88
KCTD3	613272	70	94	92	86
KIAA0556	616650	181	100	100	100
KIAA0586	610178	62	96	86	75
KIAA0753	617112	101	100	97	95
KIF14	611279	60	91	81	73
KIF7	611254	224	100	100	98
LBR	600024	72	96	94	86
LCA5	611408	64	95	94	88
LEFTY2	601877	342	100	100	100
LRRC56	618227	212	100	100	100
LRRC6	614930	73	93	91	86
LZTFL1	606568	43	84	76	70
MAPKBP1	616786	198	100	100	100
MCIDAS	614086	211	100	100	100
MKKS	604896	251	100	100	100
MKS1	609883	157	100	100	100
MMP21	608416	151	100	100	99
NCAPG2	608532	81	97	91	85
NEK1	604588	53	92	81	70
NEK10	618726	76	97	89	82
NEK8	609799	274	100	100	100

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NME5	603575	62	90	85	81
NME8	607421	45	87	74	62
NODAL	601265	262	100	100	100
NPHP1	607100	89	97	96	89
NPHP3	608002	89	94	89	87
NPHP4	607215	258	100	100	100
OCRL	300535	75	100	98	91
OFD1	300170	51	97	80	64
PDE6D	602676	118	100	97	84
PIBF1	607532	25	91	61	33
PIH1D3	300933	42	100	98	81
PIK3C2A	603601	65	98	92	84
PKD1	601313	252	97	97	97
PKD2	173910	102	100	95	88
PKHD1	606702	136	100	100	99
POC1A	614783	216	100	100	100
RPGRIP1	605446	119	100	99	97
RPGRIP1L	610937	80	95	91	83
RSPH1	609314	91	100	100	98
RSPH3	615876	191	100	99	94
RSPH4A	612647	131	100	100	97
RSPH9	612648	215	100	100	100
SCLT1	611399	43	89	76	62
SDCCAG8	613524	66	100	91	78
SPAG1	603395	59	93	80	68
SPATA7	609868	114	88	81	77
SUFU	607035	206	100	100	100
TBC1D32	615867	44	90	76	59
TCTEX1D2	617353	146	100	100	88
TCTN1	609863	117	98	97	95
TCTN2	613846	108	100	95	93
TCTN3	613847	126	100	100	98
TMEM107	616183	136	100	100	100
TMEM138	614459	110	100	100	100
TMEM216	613277	142	100	100	100
TMEM231	614949	271	100	100	98
TMEM237	614423	77	98	98	95
TMEM260	617449	77	100	93	86
TMEM67	609884	61	90	74	62
TRAF3IP1	607380	80	99	90	81
TRIM32	602290	222	100	100	100
TTBK2	611695	141	100	100	100
TTC12	610732	116	100	98	95

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TTC21B	612014	63	99	95	83
TTC25	617095	121	100	100	97
TTC26	617453	79	100	99	94
TTC8	608132	104	96	91	86
TULP1	602280	172	100	97	92
WDPCP	613580	87	98	91	82
WDR19	608151	75	100	96	92
WDR34	613363	177	100	100	100
WDR35	613602	75	97	93	87
WDR60	615462	85	99	95	88
XPNPEP3	613553	157	100	100	99
ZIC3	300265	137	100	100	99
ZMYND10	607070	232	100	100	100
ZNF423	604557	316	100	100	100

- OMIM release used: 8-9-2019
- 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