

## Supplementary tables

Supplementary Table 1. Summary of genetic diagnosis rates of various ciliopathies using different next generation sequencing approaches.

Patient group	Sequencing approach	Genetic diagnosis rate	Year	Reference
10 families with NPHP	homozygosity mapping + WES	70%	2014	(1)
Syndromic ciliopathies	Gene panel, SNP genotyping, targeted sequencing of candidate genes	62%	2015	(2)
375 families with JBTS	Gene panel	32%	2015	(3)
Ciliopathy patients in a large rare disease cohort	WES	44%	2015	(4)
79 suspected NPHP cases	WES	40.5%	2016	(5)
43 patients with NPHP who had had the 5 most common NPHP genes excluded as a cause of their disease	Gene panel	16.3%	2016	(6)
26 patients with JBTS or MKS	WES with split read mapping	46%	2016	(7)
6 BBS patients	WES	67%	2017	(8)
100 families with JBTS	Gene panel sequencing + WES	94%	2017	(9)
56 families in whom PCD was suspected	WES	68%	2020	(10)
125 families with ciliopathies	WGS	87%	2020	(11)
29 families with skeletal ciliopathies	WGS + structural variant screening + RNA analyses	90%	2021	(12)

Supplementary Table 2. Diagnostic grade “green” genes in Rare Multisystem Ciliopathy Disorders PanelApp panel V1.139

Gene name	Mode of inheritance	Associated phenotypes	Ensembl Id
<b>AHI1</b>	Biallelic	JBTS	ENSG00000135541
<b>ALMS1</b>	Biallelic	BBS, Alström	ENSG00000116127
<b>ANKS6</b>	Biallelic	PKD, NPHP	ENSG00000165138
<b>ARL13B</b>	Biallelic	JBTS	ENSG00000169379
<b>ARL6</b>	Biallelic	BBS	ENSG00000113966
<b>ARMC9</b>	Biallelic	JBTS	ENSG00000135931
<b>B9D2</b>	Biallelic	JBTS, MKS	ENSG00000123810
<b>BBS1</b>	Biallelic	BBS	ENSG00000174483
<b>BBS10</b>	Biallelic	BBS	ENSG00000179941

<b>BBS12</b>	Biallelic	BBS	ENSG00000181004
<b>BBS2</b>	Biallelic	BBS	ENSG00000125124
<b>BBS4</b>	Biallelic	BBS	ENSG00000140463
<b>BBS5</b>	Biallelic	BBS	ENSG00000163093
<b>BBS7</b>	Biallelic	BBS	ENSG00000138686
<b>BBS9</b>	Biallelic	BBS	ENSG00000122507
<b>C21orf2</b>	Biallelic	JATD, Spondylometaphyseal dysplasia, IRD	ENSG00000160226
<b>C2CD3</b>	Biallelic	SRPS, JATD, OFD	ENSG00000168014
<b>C5orf42</b>	Biallelic	JBTS, OFD	ENSG00000197603
<b>CC2D2A</b>	Biallelic	JBTS, MKS, COACH	ENSG00000048342
<b>CENPF</b>	Biallelic	Stromme	ENSG00000117724
<b>CEP104</b>	Biallelic	JBTS	ENSG00000116198
<b>CEP120</b>	Biallelic	SRTD, CED, JATD	ENSG00000168944
<b>CEP164</b>	Biallelic	NPHP, SLS	ENSG00000110274
<b>CEP290</b>	Biallelic	JBTS, MKS, COACH, SLS	ENSG00000198707
<b>CEP41</b>	Biallelic	JBTS	ENSG00000106477
<b>CEP83</b>	Biallelic	NPHP	ENSG00000173588
<b>CRB2</b>	Biallelic	PKD with ventriculomegaly	ENSG00000148204
<b>CSPP1</b>	Biallelic	JBTS, MKS	ENSG00000104218
<b>DDX59</b>	Biallelic	OFD	ENSG00000118197
<b>DHCR7</b>	Biallelic	SLO	ENSG00000172893
<b>DYNC2H1</b>	Biallelic	SRTD, CED, JATD	ENSG00000187240
<b>DYNC2LI1</b>	Biallelic	SRTD	ENSG00000138036
<b>EVC</b>	Biallelic	EVC, WAD	ENSG00000072840
<b>EVC2</b>	Biallelic	EVC, WAD	ENSG00000173040
<b>GLI3</b>	Monoallelic	JBTS, SLS	ENSG00000106571
<b>HNF1B</b>	Monoallelic	PKD, NPHP	ENSG00000275410
<b>HYLS1</b>	Biallelic	JBTS, Hydroletharus syndrome	ENSG00000198331
<b>ICK</b>	Biallelic	Endocrine-cerebro-osteodysplasia	ENSG00000112144
<b>IFT122</b>	Biallelic	CED	ENSG00000163913
<b>IFT140</b>	Biallelic	SRTD, JATD, Mainzer-Saldino	ENSG00000187535
<b>IFT172</b>	Biallelic	RP, SRTD, JATD, Mainzer-Saldino, SRTD	ENSG00000138002
<b>IFT27</b>	Biallelic	? BBS	ENSG00000100360
<b>IFT43</b>	Biallelic	SRTD, CED, Sensenbrennar syndrome	ENSG00000119650
<b>IFT52</b>	Biallelic	SRTD	ENSG00000101052
<b>IFT74</b>	Biallelic	? BBS	ENSG00000096872
<b>IFT80</b>	Biallelic	SRTD, JATD	ENSG00000068885
<b>INPP5E</b>	Biallelic	JBTS	ENSG00000148384
<b>INVS</b>	Biallelic	NPHP, SLS	ENSG00000119509
<b>IQCB1</b>	Biallelic	SLS	ENSG00000173226

<b>KIAA0586</b>	Biallelic	JBTS, SRTD	ENSG00000100578
<b>KIAA0753</b>	Biallelic	OFD, SRTD, JBTS	ENSG00000198920
<b>KIF7</b>	Biallelic	JBTS, Acrocallosal syndrome	ENSG00000166813
<b>LZTFL1</b>	Biallelic	BBS	ENSG00000163818
<b>MAPKBP1</b>	Biallelic	NPHP	ENSG00000137802
<b>MKKS</b>	Biallelic	BBS	ENSG00000125863
<b>MKS1</b>	Biallelic	MKS, BBS, JBTS	ENSG00000011143
<b>NEK1</b>	Biallelic	SRTD	ENSG00000137601
<b>NEK8</b>	Biallelic	NPHP	ENSG00000160602
<b>NPHP1</b>	Biallelic	NPHP, JBTS, SLS	ENSG00000144061
<b>NPHP3</b>	Biallelic	MKS, SLS, NPHP	ENSG00000113971
<b>NPHP4</b>	Biallelic	NPHP, SLS	ENSG00000131697
<b>OFD1</b>	X-linked	JBTS, OFD	ENSG00000046651
<b>PIBF1</b>	Biallelic	JBTS	ENSG00000083535
<b>PKD1</b>	Monoallelic and biallelic	PKD	ENSG00000008710
<b>PKD2</b>	Monoallelic	PKD	ENSG00000118762
<b>PKHD1</b>	Biallelic	Polycystic kidney and hepatic disease	ENSG00000170927
<b>PMM2</b>	Biallelic	Congenital disorder of glycosylation	ENSG00000140650
<b>RPGRIP1L</b>	Biallelic	JBTS, MKS	ENSG00000103494
<b>SBDS</b>	Biallelic	Skeletal ciliopathies	ENSG00000126524
<b>SCLT1</b>	Biallelic	OFD, SLS	ENSG00000151466
<b>SDCCAG8</b>	Biallelic	BBS, SLS	ENSG00000054282
<b>TCTEX1D2</b>	Biallelic	SRTD, JATD	ENSG00000213123
<b>TCTN1</b>	Biallelic	JBTS	ENSG00000204852
<b>TCTN2</b>	Biallelic	JBTS, MKS	ENSG00000168778
<b>TCTN3</b>	Biallelic	OFD, JBTS, MKS, Mohr-Majewski syndrome	ENSG00000119977
<b>TMEM107</b>	Biallelic	MKS, OFD, ? JBTS	ENSG00000179029
<b>TMEM138</b>	Biallelic	JBTS	ENSG00000149483
<b>TMEM216</b>	Biallelic	JBTS, MKS	ENSG00000187049
<b>TMEM231</b>	Biallelic	JBTS, MKS	ENSG00000205084
<b>TMEM237</b>	Biallelic	JBTS	ENSG00000155755
<b>TMEM67</b>	Biallelic	JBTS, MKS, COACH, NPHP, Senior-Boichis syndrome, ? BBS	ENSG00000164953
<b>TRAF3IP1</b>	Biallelic	SLS	ENSG00000204104
<b>TTC21B</b>	Biallelic	NPHP, SRTD, JATD	ENSG00000123607
<b>TTC8</b>	Biallelic	BBS	ENSG00000165533
<b>TXNDC15</b>	Biallelic	MKS	ENSG00000113621
<b>VPS13B</b>	Biallelic	Cohen syndrome	ENSG00000132549
<b>WDPCP</b>	Biallelic	MKS, ?BBS	ENSG00000143951
<b>WDR19</b>	Biallelic	CED, SRTD, JATD, NPHP, SLS	ENSG00000157796

<b>WDR34</b>	Biallelic	SRTD, JATD	ENSG00000119333
<b>WDR35</b>	Biallelic	CED, SRTD	ENSG00000118965
<b>WDR60</b>	Biallelic	SRTD, JATD	ENSG00000126870
<b>ZSWIM6</b>	Monoallelic	Acromelic frontonasal dysostosis	ENSG00000130449
<b>ISCA-37405-Loss</b>	Biallelic	JBTS, NPHP	
<b>ISCA-37432-Loss</b>	Monoallelic	Renal cysts and diabetes syndrome, Autism Spectrum Disorder, Mayer-Rokitansky-Kuster-Hauser syndrome	

**Abbreviations:** BBS: Bardet Biedl syndrome; CED: Cranioectodermal dysplasia; COACH: cerebellar vermis hypo/aplasia, oligophrenia, ataxia, ocular coloboma, and hepatic fibrosis; EVC: Ellis-Van-Creveld syndrome; IRD: Inherited retinal dystrophy; JATD: Jeune asphyxiating thoracic dystrophy; JBTS: Joubert syndrome; LCA: Leber's congenital amaurosis; MKS: Meckel Gruber Syndrome; NPHP: nephronophthisis; PKD: Polycystic kidney disease; RP: Retinitis Pigmentosa; SLO: Smith Lemli Optiz syndrome; SLS: Senior Loken syndrome; SRPS: Short rib polydactyly syndrome; SRTD: Short rib thoracic dystrophy; WAD: Weyers acrofacial dysostosis

Supplementary Table 2. Candidate gene list provided alongside Rare Multisystem Super Ciliopathy panel v4.91 for Extract\_hets Python script.

Gene symbol	Source	Ensembl Id
ACVR2B	PanelApp RMCD red gene	ENSG00000114739
ADAMTS10	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000142303
ADAMTS9	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000163638
ADCY3	On SCGS V1 list	ENSG00000138031
ADGRV1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000164199
AIPL1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000129221
AK7	On SCGS V1 list	ENSG00000140057
AK8	On SCGS V1 list	ENSG00000165695
ARF4	On SCGS V1 list	ENSG00000168374
ARL3	On SCGS V1 list	ENSG00000138175
ARMC4	PanelApp RMCD red gene	ENSG00000169126
ASAP1	On SCGS V1 list	ENSG00000153317
ATXN10	PanelApp RMCD red gene + SCGS V1 list	ENSG00000130638
B9D1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000108641
BBIP1	PanelApp RMCD red gene	ENSG00000214413
C21orf59	PanelApp RMCD red gene	ENSG00000159079
C2orf71	PanelApp RMCD red gene	ENSG00000179270
C8orf37	PanelApp RMCD red gene + SCGS V1 list	ENSG00000156172

CBY1	On SCGS V1 list	ENSG00000100211
CCDC103	PanelApp RMCD red gene + SCGS V1 list	ENSG00000167131
CCDC114	PanelApp RMCD red gene + SCGS V1 list	ENSG00000105479
CCDC151	PanelApp RMCD red gene	ENSG00000198003
CCDC28B	PanelApp RMCD red gene + SCGS V1 list	ENSG00000160050
CCDC39	PanelApp RMCD red gene + SCGS V1 list	ENSG00000145075
CCDC40	PanelApp RMCD red gene + SCGS V1 list	ENSG00000141519
CCDC65	PanelApp RMCD red gene	ENSG00000139537
CCNO	PanelApp RMCD red gene	ENSG00000152669
CCP110	On SCGS V1 list	ENSG00000103540
CDH23	PanelApp RMCD red gene + SCGS V1 list	ENSG00000107736
CENPJ	On SCGS V1 list	ENSG00000151849
CEP131	On SCGS V1 list	ENSG00000141577
CEP135	On SCGS V1 list	ENSG00000174799
CEP250	On SCGS V1 list	ENSG00000126001
CEP72	On SCGS V1 list	ENSG00000112877
CEP89	On SCGS V1 list	ENSG00000121289
CEP97	On SCGS V1 list	ENSG00000182504
CFAP100	On SCGS V1 list	ENSG00000163885
CFAP43	PanelApp RMCD red gene	ENSG00000197748
CFAP44	PanelApp RMCD red gene	ENSG00000206530
CFAP45	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000213085
CFAP53	PanelApp RMCD red gene	ENSG00000172361
CFAP57	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000243710
CFC1	PanelApp RMCD red gene	ENSG00000136698
CFTR	PanelApp RMCD red gene	ENSG00000001626
CLDN2	On SCGS V1 list	ENSG00000165376
CLRN1	PanelApp RMCD red gene	ENSG00000163646
CLTB	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000175416
CLUAP1	On SCGS V1 list	ENSG00000103351
CNGA2	On SCGS V1 list	ENSG00000183862
CNGA4	On SCGS V1 list	ENSG00000132259
CNGB1	On SCGS V1 list	ENSG00000070729
CRB1	PanelApp RMCD red gene	ENSG00000134376
CRB3	On SCGS V1 list	ENSG00000130545
CRELD1	PanelApp RMCD red gene	ENSG00000163703
CROCC	On SCGS V1 list	ENSG00000058453
CRX	PanelApp RMCD red gene	ENSG00000105392
CTNNB1	On SCGS V1 list	ENSG00000168036

DCDC2	PanelApp RMCD red gene + SCGS V1 list	ENSG00000146038
DISC1	On SCGS V1 list	ENSG00000162946
DNAAF1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000154099
DNAAF2	PanelApp RMCD red gene + SCGS V1 list	ENSG00000165506
DNAAF3	PanelApp RMCD red gene + SCGS V1 list	ENSG00000167646
DNAAF4	PanelApp RMCD red gene	ENSG00000256061
DNAAF5	PanelApp RMCD red gene	ENSG00000164818
DNAH1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000114841
DNAH10	On SCGS V1 list	ENSG00000197653
DNAH11	PanelApp RMCD red gene + SCGS V1 list	ENSG00000105877
DNAH2	On SCGS V1 list	ENSG00000183914
DNAH5	PanelApp RMCD red gene + SCGS V1 list	ENSG00000039139
DNAH6	On SCGS V1 list	ENSG00000115423
DNAI1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000122735
DNAI2	PanelApp RMCD red gene + SCGS V1 list	ENSG00000171595
DNAJB13	PanelApp RMCD red gene	ENSG00000187726
DNAL1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000119661
DNALI1	On SCGS V1 list	ENSG00000163879
DNHD1	PanelApp RMCD red gene	ENSG00000179532
DPCD	On SCGS V1 list	ENSG00000166171
DPYSL2	On SCGS V1 list	ENSG00000092964
DRC1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000157856
DRD1	On SCGS V1 list	ENSG00000184845
DRD2	On SCGS V1 list	ENSG00000149295
DRD5	On SCGS V1 list	ENSG00000169676
DVL1	On SCGS V1 list	ENSG00000107404
DYNLT1	On SCGS V1 list	ENSG00000146425
EFHC1	On SCGS V1 list	ENSG00000096093
EXOC3	On SCGS V1 list	ENSG00000180104
EXOC3L2	PanelApp RMCD amber gene	ENSG00000283632
EXOC4	On SCGS V1 list	ENSG00000131558
EXOC5	On SCGS V1 list	ENSG00000070367
EXOC6	On SCGS V1 list	ENSG00000138190
EXOC6B	On SCGS V1 list	ENSG00000144036
EXOC8	PanelApp RMCD red gene	ENSG00000116903
EZH2	On SCGS V1 list	ENSG00000106462
FAM149B1	PanelApp RMCD amber gene	ENSG00000138286
FAM161A	On SCGS V1 list	ENSG00000170264
FBF1	On SCGS V1 list	ENSG00000188878
FLNA	On SCGS V1 list	ENSG00000196924
FOPNL	On SCGS V1 list	ENSG00000133393

FOXH1	PanelApp RMCD red gene	ENSG00000160973
FOXJ1	On SCGS V1 list	ENSG00000129654
FUZ	On SCGS V1 list	ENSG00000010361
GAS8	PanelApp RMCD red gene + SCGS V1 list	ENSG00000141013
GDF1	PanelApp RMCD red gene	ENSG00000130283
GLI1	On SCGS V1 list	ENSG00000111087
GLI2	On SCGS V1 list	ENSG00000074047
GLIS2	PanelApp RMCD red gene + SCGS V1 list	ENSG00000126603
GPR161	On SCGS V1 list	ENSG00000143147
GSK3B	On SCGS V1 list	ENSG00000082701
GUCY2D	PanelApp RMCD red gene	ENSG00000132518
HAP1	On SCGS V1 list	ENSG00000173805
HSD11B1	On SCGS V1 list	ENSG00000117594
HSPA8	On SCGS V1 list	ENSG00000109971
HSPB11	On SCGS V1 list	ENSG00000081870
HTR6	On SCGS V1 list	ENSG00000158748
HTT	On SCGS V1 list	ENSG00000197386
HYDIN	PanelApp RMCD red gene + SCGS V1 list	ENSG00000157423
IFT122	On SCGS V1 list	ENSG00000128581
IFT20	On SCGS V1 list	ENSG00000109083
IFT46	On SCGS V1 list	ENSG00000118096
IFT57	On SCGS V1 list	ENSG00000114446
IFT81	PanelApp RMCD red gene + SCGS V1 list	ENSG00000122970
IFT88	On SCGS V1 list	ENSG00000032742
IMPDH1	PanelApp RMCD red gene	ENSG00000106348
INTU	On SCGS V1 list	ENSG00000164066
JADE1	On SCGS V1 list	ENSG00000077684
KCNJ13	PanelApp RMCD red gene	ENSG00000115474
KIAA0556	PanelApp RMCD red gene	ENSG00000047578
KIF14	PanelApp RMCD red gene	ENSG00000118193
KIF17	On SCGS V1 list	ENSG00000117245
KIF19	On SCGS V1 list	ENSG00000196169
KIF24	On SCGS V1 list	ENSG00000186638
KIF27	On SCGS V1 list	ENSG00000165115
KIF3A	On SCGS V1 list	ENSG00000131437
KIF3B	On SCGS V1 list	ENSG00000101350
KIF3C	On SCGS V1 list	ENSG00000084731
KIF5B	On SCGS V1 list	ENSG00000170759
LBR	PanelApp RMCD red gene	ENSG00000143815
LCA5	PanelApp RMCD red gene + SCGS V1 list	ENSG00000135338
LEFTY2	PanelApp RMCD red gene	ENSG00000143768
LRAT	PanelApp RMCD red gene	ENSG00000121207



LRR6	PanelApp RMCD red gene + SCGS V1 list	ENSG00000129295
LRR45	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000169683
MAK	On SCGS V1 list	ENSG00000111837
MAL	On SCGS V1 list	ENSG00000172005
MAPRE1	On SCGS V1 list	ENSG00000101367
MCHR1	On SCGS V1 list	ENSG00000128285
MCIDAS	PanelApp RMCD red gene	ENSG00000234602
MDM1	On SCGS V1 list	ENSG00000111554
MICAL2	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000133816
MLF1	On SCGS V1 list	ENSG00000178053
MNS1	On SCGS V1 list	ENSG00000138587
MUC1	PanelApp RMCD red gene	ENSG00000185499
MYO15A	On SCGS V1 list	ENSG00000091536
MYO7A	PanelApp RMCD red gene + SCGS V1 list	ENSG00000137474
NEK2	On SCGS V1 list	ENSG00000117650
NEK4	On SCGS V1 list	ENSG00000114904
NGFR	On SCGS V1 list	ENSG00000064300
NIN	On SCGS V1 list	ENSG00000100503
NINL	On SCGS V1 list	ENSG00000101004
NKX2-5	PanelApp RMCD red gene	ENSG00000183072
NME5	On SCGS V1 list	ENSG00000112981
NME7	On SCGS V1 list	ENSG00000143156
NME8	PanelApp RMCD red gene + SCGS V1 list	ENSG00000086288
NODAL	PanelApp RMCD red gene	ENSG00000156574
NOTO	On SCGS V1 list	ENSG00000214513
NUP214	On SCGS V1 list	ENSG00000126883
NUP35	On SCGS V1 list	ENSG00000163002
NUP37	On SCGS V1 list	ENSG00000075188
NUP62	On SCGS V1 list	ENSG00000213024
NUP93	On SCGS V1 list	ENSG00000102900
OCRL	PanelApp RMCD red gene + SCGS V1 list	ENSG00000122126
ODF2	On SCGS V1 list	ENSG00000136811
ORC1	On SCGS V1 list	ENSG00000085840
PACRG	On SCGS V1 list	ENSG00000112530
PAFAH1B1	On SCGS V1 list	ENSG00000007168
PARD3	On SCGS V1 list	ENSG00000148498
PARD6A	On SCGS V1 list	ENSG00000102981
PCDH15	PanelApp RMCD red gene + SCGS V1 list	ENSG00000150275
PCM1	On SCGS V1 list	ENSG00000078674
PDE6D	PanelApp RMCD red gene + SCGS V1 list	ENSG00000156973



PDZD7	On SCGS V1 list	ENSG00000186862
PKD1L1	On SCGS V1 list	ENSG00000158683
PLK1	On SCGS V1 list	ENSG00000166851
POC1A	PanelApp RMCD red gene + SCGS V1 list	ENSG00000164087
POC1B	PanelApp RMCD amber gene	ENSG00000139323
PPP3CC	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000120910
PRKCSH	PanelApp RMCD red gene	ENSG00000130175
PTCH1	On SCGS V1 list	ENSG00000185920
PTPDC1	On SCGS V1 list	ENSG00000158079
RAB11A	On SCGS V1 list	ENSG00000103769
RAB11FIP3	On SCGS V1 list	ENSG00000090565
RAB17	On SCGS V1 list	ENSG00000124839
RAB23	On SCGS V1 list	ENSG00000112210
RAB3IP	On SCGS V1 list	ENSG00000127328
RAB8A	On SCGS V1 list	ENSG00000167461
RAN	On SCGS V1 list	ENSG00000132341
RANBP1	On SCGS V1 list	ENSG00000099901
RD3	PanelApp RMCD red gene	ENSG00000198570
RDH12	PanelApp RMCD red gene	ENSG00000139988
RFX3	On SCGS V1 list	ENSG00000080298
RILPL1	On SCGS V1 list	ENSG00000188026
RILPL2	On SCGS V1 list	ENSG00000150977
ROPN1L	On SCGS V1 list	ENSG00000145491
RP1	On SCGS V1 list	ENSG00000104237
RP2	On SCGS V1 list	ENSG00000102218
RPE65	PanelApp RMCD red gene	ENSG00000116745
RPGR	PanelApp RMCD red gene + SCGS V1 list	ENSG00000156313
RPGRIP1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000092200
RSPH1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000160188
RSPH3	On SCGS V1 list	ENSG00000130363
RSPH4A	PanelApp RMCD red gene + SCGS V1 list	ENSG00000111834
RSPH9	PanelApp RMCD red gene + SCGS V1 list	ENSG00000172426
RTTN	On SCGS V1 list	ENSG00000176225
SASS6	On SCGS V1 list	ENSG00000156876
SCNN1A	PanelApp RMCD red gene	ENSG00000111319
SCNN1B	PanelApp RMCD red gene	ENSG00000168447
SCNN1G	PanelApp RMCD red gene	ENSG00000166828
SEC63	PanelApp RMCD red gene	ENSG00000025796
SEPT2	On SCGS V1 list	ENSG00000168385
SEPT7	On SCGS V1 list	ENSG00000122545
SHH	On SCGS V1 list	ENSG00000164690

SLC47A2	On SCGS V1 list	ENSG00000180638
SMCR8	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000283741
SMO	On SCGS V1 list	ENSG00000128602
SNAP25	On SCGS V1 list	ENSG00000132639
SNX10	On SCGS V1 list	ENSG00000086300
SNX17	On SCGS V1 list	ENSG00000115234
SPA17	On SCGS V1 list	ENSG00000064199
SPAG1	PanelApp RMCD red gene	ENSG00000104450
SPAG16	On SCGS V1 list	ENSG00000144451
SPAG17	On SCGS V1 list	ENSG00000155761
SPAG6	On SCGS V1 list	ENSG00000077327
SPATA7	PanelApp RMCD red gene + SCGS V1 list	ENSG00000042317
SPEF2	On SCGS V1 list	ENSG00000152582
SPTBN4	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000160460
SSNA1	On SCGS V1 list	ENSG00000176101
SSTR3	On SCGS V1 list	ENSG00000278195
STIL	On SCGS V1 list	ENSG00000123473
STK36	On SCGS V1 list	ENSG00000163482
STK38L	On SCGS V1 list	ENSG00000211455
STOML3	On SCGS V1 list	ENSG00000133115
STX3	On SCGS V1 list	ENSG00000166900
SUFU	PanelApp RMCD amber gene + SCGS V1 list	ENSG00000107882
SYNE2	On SCGS V1 list	ENSG00000054654
TAPT1	PanelApp RMCD red gene	ENSG00000169762
TBC1D30	On SCGS V1 list	ENSG00000111490
TBC1D32	PanelApp RMCD red gene	ENSG00000146350
TBC1D7	On SCGS V1 list	ENSG00000145979
TEKT2	On SCGS V1 list	ENSG00000092850
TEKT4	On SCGS V1 list	ENSG00000163060
TEKT5	On SCGS V1 list	ENSG00000153060
TEX12	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000150783
TNPO1	On SCGS V1 list	ENSG00000083312
TOPORS	PanelApp RMCD red gene + SCGS V1 list	ENSG00000197579
TPPP2	On SCGS V1 list	ENSG00000179636
TRAPPC10	On SCGS V1 list	ENSG00000160218
TRAPPC3	On SCGS V1 list	ENSG00000054116
TRAPPC9	On SCGS V1 list	ENSG00000167632
TRIM32	PanelApp RMCD red gene + SCGS V1 list	ENSG00000119401
TRIP11	On SCGS V1 list	ENSG00000100815

TSC1	PanelApp RMCD red gene	ENSG00000165699
TSC2	PanelApp RMCD red gene	ENSG00000103197
TTBK2	PanelApp RMCD red gene + SCGS V1 list	ENSG00000128881
TTC12	On SCGS V1 list	ENSG00000149292
TTC26	On SCGS V1 list	ENSG00000105948
TTC29	On SCGS V1 list	ENSG00000137473
TTC30A	On SCGS V1 list	ENSG00000197557
TTC30B	On SCGS V1 list	ENSG00000196659
TTK	On SCGS V1 list	ENSG00000112742
TTLL3	On SCGS V1 list	ENSG00000214021
TTLL6	On SCGS V1 list	ENSG00000170703
TTLL9	On SCGS V1 list	ENSG00000131044
TUBA1A	On SCGS V1 list	ENSG00000167552
TUBA1C	On SCGS V1 list	ENSG00000167553
TUBA4A	On SCGS V1 list	ENSG00000127824
TUBB2A	On SCGS V1 list	ENSG00000137267
TUBB2B	On SCGS V1 list	ENSG00000137285
TUBB3	On SCGS V1 list	ENSG00000258947
TUBB8	Ciliopathy candidate identified through local research team experimental screen(s)	ENSG00000261456
TUBE1	On SCGS V1 list	ENSG00000074935
TUBGCP2	On SCGS V1 list	ENSG00000130640
TUBGCP3	On SCGS V1 list	ENSG00000126216
TUBGCP4	On SCGS V1 list	ENSG00000137822
TUBGCP5	On SCGS V1 list	ENSG00000275835
TUBGCP6	On SCGS V1 list	ENSG00000128159
TULP1	PanelApp RMCD red gene + SCGS V1 list	ENSG00000112041
TULP3	On SCGS V1 list	ENSG00000078246
ULK4	On SCGS V1 list	ENSG00000168038
UMOD	PanelApp RMCD red gene	ENSG00000169344
USH1C	PanelApp RMCD red gene + SCGS V1 list	ENSG00000006611
USH1G	PanelApp RMCD red gene + SCGS V1 list	ENSG00000182040
USH2A	PanelApp RMCD red gene + SCGS V1 list	ENSG00000042781
VANGL2	On SCGS V1 list	ENSG00000162738
VDAC3	On SCGS V1 list	ENSG00000078668
VHL	PanelApp RMCD red gene + SCGS V1 list	ENSG00000134086
WDR63	PanelApp RMCD red gene	ENSG00000162643
WDR78	On SCGS V1 list	ENSG00000152763
WHRN	PanelApp RMCD red gene + SCGS V1 list	ENSG00000095397
XPNPEP3	PanelApp RMCD red gene + SCGS V1 list	ENSG00000196236
ZIC3	PanelApp RMCD red gene	ENSG00000156925
ZMYND10	PanelApp RMCD red gene	ENSG00000004838

ZNF423	PanelApp RMCD amber gene + SCGS V1 list	ENSG00000102935
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Abbreviations: RMCD = Rare multisystem Ciliopathy disorders, SCGS V1 = SYSCILIA gold standard list version 1

Supplementary Table 4. Detailed variant information for all variants identified amongst participants of the congenital malformations caused by ciliopathies cohort with research identified diagnoses.

Proband research number	Gene symbol	HGVSc	HGVSp	Segregation	Zygoty (presumed where segregation data unk)	Mutational mechanism	SIFT	PolyPhen	1000G AF	gnomAD AF	ClinVar	PMID	CADD Phred	SpliceAI max DS	Match for phenotype	Variant ACMG class
1	<i>CHD7</i>	NM_017780.4:c.6955C>T	NP_060250.2:p.Arg2319Cys	De Novo	Het	Mis	Delet (0)	Prob_dam (0.99)	-	-	Path	16400610	27.1		Full	5
2	<i>ALMS1</i>	NM_001378454.1:c.10772del	NP_001365383.1:p.Thr3591LysfsTer6	Pat	Comp het	SG	-	-	-	0.00005227	Path	11941369, 11941370, 17594715	23		Full	5
2	<i>ALMS1</i>	NM_001378454.1:c.11104C>T	NP_001365383.1:p.Arg3702Ter	Mat	Comp het	SG	-	-	-	0.00001205	Path	-	34		Full	5
3	<i>ARL6</i>	NM_001278293.3:c.534A>G	NP_001265222.1:p.Gln178=	Bi-parental	Hom	Syn	-	-	-	0.00000796	-	27708425	24.1	DS_DL 0.87	Full	5
3	<i>IMPG2</i>	NM_016247.4:c.3262C>T	NP_057331.2:p.Arg1088Ter	Bi-parental	Hom	SG	-	-	0.0002	0.0000358	Path, Lik_Path	-	42		Partial	4
4	<i>RPGR</i>	NM_000328.3:c.1627del	NP_000319.1:p.Asp5431IlefsTer11	Unk	XLR	FS, male	-	-	-	-	-	-	-		Partial	4
5	<i>CEP290</i>	NM_025114.4:c.5668G>T	NP_079390.3:p.Gly1890Ter	Unk	Comp het	SG	-	-	0.0002	0.00009486	Path	18414213, 26092869, 16682970, 16682973, 17564967	35		Full	5
5	<i>CEP290</i>	NM_025114.4:c.322C>T	NP_079390.3:p.Arg108Ter	Unk	Comp het	SG	-	-	-	0.00000402	Path	-	35		Full	5
6	<i>KIAA0586</i>	NM_001329943.3:c.392del	NP_001316872.1:p.Arg1311LysfsTer4	Pat	Comp het	FS	-	-	0.0024	0.00305	Path, Lik_Path	25741868, 26096313, 28552196, 26386247	33		Full	5
6	<i>KIAA0586</i>	NM_001329943.3:c.1402_1408del	NP_001316872.1:p.Pro468IlefsTer15	Mat	Comp het	FS	-	-	-	-	-	-	-		Full	5
7	<i>CRYBB1</i>	NM_001887.4:c.193G>A	NP_001878.1:p.Glu65Lys	Unk	Het	Mis	Delet (0)	Prob_dam (0.979)	-	-	-	-	29.2		Partial	3
7	<i>OFD1</i>	NM_003611.3:c.306del	NP_003602.1:p.Glu103LysfsTer42	Unk	XLD	FS, female	-	-	-	-	-	-	-		Full	4

7	<i>PKD1</i>	NM_001009 944.3:c.5890 C>T	NP_00100 9944.3:p.A rg1964Cys	Unk	Het	Mis	Delet (0.03)	poss_dam (0.772)	-	0.00001316	-	-	25.5		Partial	3
8	<i>PRPF8</i>	NM_006445. 4:c.5804G>A	NP_00643 6.3:p.Arg1 935His	De Novo	Het	Mis	Delet (0)	Prob_dam (1)	-	-	Path, Lik_Path	-	28.3		Full	4
9	<i>CEP152</i>	NM_001194 998.2:c.2041 C>T	NP_00118 1927.1:p.H is681Tyr	Pat	Comp het	Mis	Delet (0)	Prob_dam (0.999)	0.0002	0.0001603	VUS	-	25.1		Partial	3
9	<i>CEP152</i>	NM_001194 998.2:c.1499 A>T	NP_00118 1927.1:p.G lu500Val	Mat	Comp het	Mis	Delet (0.01)	Ben (0.159)	0.0002	-	-	-	23.2		Partial	3
10	<i>CEP290</i>	NM_025114. 4:c.5932C>T	NP_07939 0.3:p.Arg1 978Ter	Bi-parental	Hom	SG	-	-	-	0.00001212	Path	26092869	42		Full	5
11	<i>MYCN</i>	NM_005378. 6:c.1006del	NP_00536 9.2:p.Ser3 36LeufsTer 15	De Novo	Het	FS	-	-	-	-	-	-	-		Full	5
12	<i>ARMC9</i>	NM_001271 466.4:c.879 G>A	NP_00125 8395.2:p.T hr293=	Bi-parental	Hom	Syn	-	-	-	0.00000806 5	Path, Lik_Path	29159890, 17576681, 9536098	26.8	DS_DL 0.9	Full	5
13	<i>TUBA1A</i>	NM_006009. 4:c.641G>T	NP_00600 0.2:p.Arg2 14Leu	De Novo	Het	Mis	Delet_l ow conf (0.03)	Ben (0.023)	-	-	Path, Lik_Path	25741868	23.5		Poor	5
14	<i>WDR19</i>	NM_025132. 4:c.1630_16 39del	NP_07940 8.3:p.Val5 44LeufsTer 72	Mat	Comp het	Spl A	-	-	-	-	-	-	-		Full	5
14	<i>WDR19</i>	NM_025132. 4:c.817A>G	NP_07940 8.3:p.Asn2 73Asp	Pat	Comp het	Mis	Tol (0.28)	Ben (0.003)	-	0.00001629	VUS, Path	29068549	14.46		Full	5
15	<i>RHO</i>	NM_000539. 3:c.133T>C	NP_00053 0.1:p.Phe4 5Leu	Unk	Het	Mis	Tol (0.53)	Prob_dam (0.979)	0.0002	0.00002386	Path	1862076	23.6		Full	5
16	<i>STAG1</i>	NM_005862. 3:c.1033G>A	NP_00585 3.2:p.Glu3 45Lys	De Novo	Het	Mis	Delet (0)	Prob_dam (0.954)	-	-	-	-	28.6		Full	3
17	<i>BBS1</i>	NM_024649. 5:c.1169T>G	NP_07892 5.3:p.Met3 90Arg	Bi-parental	Hom	Mis	Delet (0)	Ben (0.347)	0.001	0.001512	Path, Lik_Path	15 PMIDs	26.2		Full	5
18	<i>RERE</i>	NM_001042 681.2:c.4286 A>T	NP_00103 6146.1:p.H is1429Leu	De Novo	Het	Mis	Delet_l ow conf (0)	Prob_dam (0.936)	-	-	-	-	28		Full	4
19	<i>ALMS1</i>	NM_001378 454.1:c.4681 _4687dup	NP_00136 5383.1:p.II e1563Asnf sTer20	Bi-parental	Hom	FS	-	-	-	-	-	-	-		Full	5
20	<i>BBS2</i>	NM_031885. 5:c.471+1G> C		Unk	Comp het	Spl d	-	-	-	-	-	-	33	DS_DL 0.95	Full	4
20	<i>BBS2</i>	NM_031885. 5:c.646C>T	NP_11409 1.4:p.Arg2 16Ter	Unk	Comp het	SG	-	-	-	0.00001194	Path, Lik_Path	11567139	37		Full	4

21	KAT6A	NM_006766. 5:c.1121C>T	NP_00675 7.2:p.Ser3 74Leu	Unk	Het	Mis	Delet (0)	Prob_dam (0.966)	-	0.00006369	-	-	28.4		Partial	3
21	LAMA1	NM_005559. 4:c.3397C>T	NP_00555 0.2:p.Arg1 133Ter	Unk	Comp het	SG	-	-	-	0.00001997	Path	-	38		Full	4
21	LAMA1	NM_005559. 4:c.281A>G	NP_00555 0.2:p.Gln9 4Arg	Unk	Comp het	Mis	Delet (0)	Prob_dam (1)	-	-	-	-	26.4		Full	3
22	MKKS	NM_170784. 2:c.1017_10 18del	NP_74075 4.1:p.Ile33 9MetfsTer 3	Bi-parental	Hom	FS	-	-	-	-	-	-	-		Full	5
23	CEP290	NM_025114. 4:c.5668G>T	NP_07939 0.3:p.Gly1 890Ter	Pat	Comp het	SG	-	-	0.0002	0.00009486	Path	18414213, 26092869, 16682970, 16682973, 17564967	36		Full	5
23	CEP290	NM_025114. 4:c.104T>G	NP_07939 0.3:p.Val3 5Gly	Mat	Comp het	Mis	Delet (0)	Prob_dam (0.943)	-	-	-	-	33		Full	3
25	RAI1	NM_030665. 4:c.2479C>T	NP_10959 0.3:p.Gln8 27Ter	De Novo	Het	SG	-	-	-	-	-	-	35		Full	5
26	PROM1	NM_006017. 3:c.1354dup	NP_00600 8.1:p.Tyr4 52LeufsTer 13	Unk	Hom	FS	-	-	-	0.0002195	Path, Lik_Path	-	32		Full	5
27	SETD2	NM_014159. 7:c.5218C>T	NP_05487 8.5:p.Arg1 740Trp	1 parent's unk, absent from other	Het	Mis	Delet (0)	Prob_dam (0.998)	-	-	Path, Lik_Path, VUS	-	32		Full	5
28	OPA1	NM_130837. 3:c.2678del	NP_57085 0.2:p.His89 3LeufsTer9	1 parent's unk, absent from other	Het	FS	-	-	-	-	-	-	-		Partial	5
29	ALMS1	NM_015120. 4:c.11881dup	NP_05593 5.4:p.Ser3 961PhefsT er12	Unk	Comp het	FS	-	-	-	-	-	-	-		Full	5
29	ALMS1	NM_015120. 4:c.1241- 81_1252deli nsCCTGCAG GCCCTCCAC ATATGCTAC AAAATA	-	Unk	Comp het	Spl A	-	-	-	-	-	-	-		Full	4
30	PHIP	NM_017934. 7:c.3202delA insTACCTG	NP_06040 4.4:p.Ile10 68AsnfsTer 3	Unk	Het	FS	-	-	-	-	-	-	-		Full	5
31	RAB28	NM_001017 979.3:c.58dup	NP_00101 7979.1:p.A sp20GlyfsT er62	Unk	Hom	FS	-	-	-	0.00000450 4	-	-	33		Full	5
32	ALMS1	GMC questionnaire reports		Unk	Comp het	Exon del	-	-	-	-	-	-	-		False positive	False positive



		exon 11 Deletion															
32	ALMS1	NM_015120.4:c.9001C>T	NP_055935.4:p.Gln3001Ter	Unk	Comp het	SG	-	-	-	0.000004008	-	-	36		Full	5	
34	POLA1	NM_001330360.2:c.460G>T	NP_001317289.1:p.Asp154Tyr	Mat	XLR	Mis, male	Delet (0)	Prob_dam (0.935)	-	0.0001049	-	-	32		Partial	3	
41	CSPP1	NM_001382391.1:c.2968+5G>A		Mat	Comp het	Spl region	-	-	-	-	VUS	-	-	DS_DL 0.79	Full	3	
41	CSPP1	GrCh38: Chr8:67136672_67139048del		Pat	Comp het	2.7kb del incl exon 15									Full	5	
42	PIBF1	NM_006346.4:c.1205T>C	NP_006337.2:p.Met402Thr	Unk	Comp het	Mis	Tol (0.09)	Ben (0.157)	-	-	-	-	22.8		Full	3	
42	PIBF1	GrCh38: chr13:g.72783352_72796671del		Unk	Comp het	13.3kb del incl exons 2-4									Full	5	
48	LRRC45	NM_144999.4:c.1074_1075insTG	NP_659436.1:p.Leu59CysfsTer19	Bi-parental	Hom	FS	-	-	-	-	-	-	-		Candidate gene	Candidate gene	
56	BBS9	NM_198428.3:c.1028G>A	NP_940820.1:p.Gly343Glu	1 parent's unk, present in other	Hom	Mis	Delet (0)	Prob_dam (1)	-	-	-	-	25.9		Full	3	
61	WT1	NM_024426.6:c.1400G>A	NP_077744.4:p.Arg467Gln	Unk	Het	Mis	Delet_low conf (0.02)	Poss_dam (0.767)	-	-	Path	1302008	29.9		Full	5	
67	ALMS1	NM_015120.4:c.1612C>G	NP_055935.4:p.Leu538Val	Pat	Comp het	Mis	Tol_low conf (0.17)	Ben (0.009)	-	0.0003289	lik_ben, VUS	-	0.003		Full	3	
67	ALMS1	NM_015120.4:c.9613A>G	NP_055935.4:p.Ile3205Val	Mat	Comp het	Mis	Delet (0.03)	Prob_dam (0.997)	-	0.000008021	VUS	-	23		Full	3	
69	BBS1	NM_024649.5:c.1169T>G	NP_078925.3:p.Met390Arg	Mat	Comp het	Mis	Delet (0)	Ben (0.347)	0.001	0.001512	Path, Lik_Path	15 PMIDs	26.2		Full	5	
69	BBS1	NM_024649.5:c.592-1333_831-449del		Pat	Comp het	4.7kb del incl exons 8 and 9									Full	5	
71	BCORL1	NM_001379451.1:c.3463C>A	NP_001366380.1:p.Pro1155Thr	Mat	XLR	Mis, male	Tol (0.12)	Poss_dam (0.557)	-	0.00003468	-	-	21.2		Partial	3	
72	CHD4	NM_001273.5:c.3225G>T	NP_001264.2:p.Met1075Ile	1 parent's unk, absent from other	Het	Mis	Delet (0)	Ben (0.358)	-	-	-	-	26		Full	3	
75	BBS4	NM_033028.5:c.642+3A>T		1 parent's unk, present in other	Hom	Spl region	-	-	-	-	-	-	22.5	DS_DL 0.82	Data missing	3	

76	<i>BBS10</i>	NM_024685.4:c.1790G>A	NP_078961.3:p.Gly597Asp	1 parent's unk, present in other	Hom	Mis	Delet (0)	Prob_dam (0.999)	-	-	-	-	25.8		Full	3
82	<i>IDUA</i>	NM_000203.5:c.1469T>C	NP_000194.2:p.Leu490Pro	Unk	Comp het	Mis	Delet (0.02)	Ben (0.086)	-	0.00004681	Path	23757202, 7550232	18.91		Full	5
82	<i>IDUA</i>	NM_000203.5:c.406G>A	NP_000194.2:p.Ala136Thr	Unk	Comp het	Mis	Delet (0.01)	Ben (0.085)	-	0.0001039	VUS	-	20.7		Full	3

Variants are described using Human Genome Variation Society (HGVS) nomenclature with the RefSeq transcript selected (or longest transcript if RefSeq not available). They are accompanied by the GEL tier, zygosity (presumed where segregation data unavailable), mutational mechanism, ACMG classification, match to phenotype and assigned diagnostic confidence.

**Abbreviations:** mat = maternal, pat = paternal, unk = unknown, AF = allele frequency, het = heterozygous, comp het = compound heterozygous, XLR = X-linked recessive, XLD = X-linked dominant, hom = homozygous, spl = splice, Spl D = splice donor, Spl A = splice acceptor, Syn = synonymous, SG = stop gain, FS = frameshift, del = deletion, kb = kilobase, low\_conf = low confidence, Tol = Tolerated, delet = deleterious, Prob\_dam = probably damaging, ben = benign, poss\_dam = possibly damaging, incl = including, Path = Pathogenic, Lik\_Path = likely Pathogenic, VUS = variant of uncertain significance, lik\_ben = likely benign, PMID = PubMed identifier, CADD = Combined Annotation Dependent Depletion (CADD), DS = delta score, DS\_DL = delta score donor loss,

**ACMG classifications:** Class 5 = pathogenic, Class 4 = likely pathogenic, Class 3 = VUS

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