Table S1. PCD genes analysed from the multi-gene panel.

Gene ID	Transmission electron microscopy (TEM) defects	Genomic Interval	# exons
ARMC4	ODA	chr10:28089736-28284096	22
CFAP298/C21ORF59	ODA+IDA	chr21:33951107-33984578	10
CCDC103	ODA+IDA	chr17:42978342-42980210	3
CCDC114	ODA	chr19:48800208-48822053	13
CCDC151	ODA	chr19:11531478-11546617	16
CCDC39	MT disorganised+IDA	chr3:180331609-180466094	24
CCDC40	MT disorganised+IDA	chr17:78010437-78073599	24
CCDC65	MT disorganised/sub-TEM defect	chr12:49298095-49325285	9
CCNO	Reduced cilia numbers	chr5:54527178-54529376	3
DNAAF1	ODA+IDA	chr16:84179021-84212037	15
DNAAF2	ODA+IDA	chr14:50092235-50101892	3
DNAAF3	ODA+IDA	chr19:55670405-55678041	12
DNAAF5	ODA+IDA	chr7:766333-825315	13
DNAH11	Normal (sub-TEM defect)	chr7:21582839-21940897	85
DNAH5	ODA	chr5:13692068-13944572	79
DNAI1	ODA	chr9:34458979-34520779	21
DNAI2	ODA	chr17:72277761-72310380	12
DNAJB13	CP	chr11:73662090-73681184	8
DNAL1	ODA	chr14:74111718-74162670	8
DRC1	MT disorganised/sub-TEM defect	chr2:26624833-26679410	19
DYX1C1	ODA+IDA	chr15:55710208-55790552	10
GAS8	Normal (sub-TEM defect)	chr16:90089105-90109778	11
HYDIN	CP/sub-TEM defect	chr16:70841458-71264615	90
LRRC6	ODA+IDA	chr8:133584529-133687764	15
MCIDAS	Reduced cilia numbers	chr5:54516169-54522991	7
NME8	ODA	chr7:37889844-37936719	15
OFD1	Not published	chrX:13753166-13787252	24
PIH1D3	ODA+IDA	chrX:106456081-106486553	6
RPGR	Mixed	chrX:38128854-38186645	20
RSPH1	CP	chr21:43892903-43916321	9
RSPH3	CP	chr6:159398545-159421033	8
RSPH4A	CP	chr6:116937762-116953629	6
RSPH9	CP	chr6:43612811-43640212	7
SPAG1	ODA+IDA	chr8:101174484-101253275	18
STK36	CP/sub-TEM defect	chr2:219537528-219566757	28
TTC25	ODA	chr17:40086952-40117526	11
ZMYND10	ODA+IDA	chr3:50378816-50383035	12

Gene names according to HUGO Gene Nomenclature Committee. Associated TEM defects: cilia outer dynein arms (ODA), inner dynein arms (IDA), peripheral microtubules (MT), central pair microtubules (CP); sub-TEM defect, indicates subtle structural defects generally not detectable using conventional TEM analysis. Note that *CFAP300*, *DNAH9*, *GAS2L2*, *SPEF2*, *LRRC56*, *MNS1*, *FOXJ1*, *DNAH1*, *DNAH6* were all also on the panel, these have become associated with PCD in the interim study period, hence they are not included to this table[1-9].

Table S2. NGS multi-gene panel design and mean coverage of targets

	651 gene panel version	**321 gene panel version**
Genome build	(Homo sapiens, hg19, GRCh37, February 2009)	
Target genes	651	321
Region	Coding exons	
Region extension	25 bases from 5' end and 25 bases from 3' end	
Region size (Megabase pairs)	2.182 Mbp	1.265 Mbp
Total probes	50,630	41,785
Total probe size (Megabase pairs)	3.536 Mbp	1.757 Mbp
Mean coverage of targets	329.08	262.5

Table S3. Details of all the 167 variants detected in 128 PCD families, in known PCD genes.

Study ID	TEM	Ethnicity Category	Cons	Gene	Genomic Coordinates (GRCh37)	Genotype	Impact	Transcript	Protein	cDNA change	Protein change	2018 HGMD ref	ExAC MAF	almena MAF	BinB MAF	Segregation	Path Class	PVS	PS	PM	PP
PCD-G060	ODA	European	No	*ARMC4*	Chr10:28257856	Comp Het	Frameshift	NM_001290020.1	NP_001276949.1	c.1233_1234delinsT	p.Leu411Phefs*48	NR	Absent	Absent	Absent	Fa carrier	5	PVS1		PM2	PP1, PP4
					Chr10:28229509		Nonsense	NM_001290020.1	NP_001276949.1	c.1969C>T	p.Gln657*	22	Absent	Absent	Absent	Mo carrier	5	PVS1	PS3	PM2	PP4
PCD-G067	ODA	Other	Unk	*ARMC4*	Chr10:28250600	Homoz	Nonsense	NM_001290020.1	NP_001276949.1	c.1283C>G	p.Ser428*	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G056	MT disorg.+IDA	European	Unk	*CCDC39*	Chr3:180372650-180372652	Homoz	Frameshift	NM_181426.1	NP_852091.1	c.830_831delCA	p.Asn276Lysfs*4	5, 7	0.00006395	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G081	MT disorg.+IDA	Arab	Yes	*CCDC39*	Chr3:180359783-180359784	Homoz	Frameshift	NM_181426.1	NP_852091.1	c.1871_1872delTA	p.Ile624Lysfs*3	10	Absent	Absent	Absent	2 cousins, Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G087	Not done	South-Asian	Unk	*CCDC39*	Chr3:180361907	Homoz	Splice donor	NM_181426.1	NP_852091.1	c.1665+1G>T	p.?	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP3, PP4
PCD-G102	MT disorg.	Arab	yes	*CCDC39*	Chr3:180359783-180359784	Homoz	Frameshift	NM_181426.1	NP_852091.1	c.1871_1872delTA	p.Ile624Lysfs*3	10	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G103	MT disorg.	Arab	yes	*CCDC39*	Chr3:180359783-180359784	Homoz	Frameshift	NM_181426.1	NP_852091.1	c.1871_1872delTA	p.Ile624Lysfs*3	10	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G106	MT disorg.	Arab	yes	*CCDC39*	Chr3:180359783-180359784	Homoz	Frameshift	NM_181426.1	NP_852091.1	c.1871_1872delTA	p.Ile624Lysfs*3	10	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G110	Not done	Arab	No	*CCDC39*	Chr3:180337130	Homoz	Nonsense	NM_181426.1	NP_852091.1	c.2182C>T	p.Gln728*	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G113	Not done	Arab	Yes	*CCDC39*	Chr3:180381653	Homoz	Splice donor	NM_181426.1	NP_852091.1	c.210+2T>C	p.?	NR	8.327E-06	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G005	MT disorg.+IDA	South-Asian	No	*CCDC40*	Chr17:78073323	Homoz	Splice site	NM_017950.3	NP_060420.2	c.3181-3C>G	p.?	NR	Absent	Absent	Absent	No parental DNA	3			PM2	PP2-4
PCD-G007	MT disorg.+IDA	European	No	*CCDC40*	Chr17:78032753	Comp Het	Frameshift	NM_017950.3	NP_060420.2	c.1414delC	p.Arg472Glyfs*3	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr17:78071119		Nonsense	NM_017950.3	NP_060420.2	c.3097A>T	p.Lys1033*	6	Absent	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	
PCD-G008	MT disorg.+IDA	European	No	*CCDC40*	Chr17:78063562	Comp Het	Splice acceptor	NM_017950.3	NP_060420.2	c.2712-1G>T	p.?	7	0.00003315	Absent	Absent	Mo normal, no Fa DNA	5	PVS1	PS3		PP4
					Chr17:78013765		Frameshift	NM_017950.3	NP_060420.2	c.248delC	p.Ala83Valfs*84	5, 8	0.0004794	Absent	Absent	Mo carrier, no Fa DNA	5	PVS1	PS3	PM2	PP4
PCD-G018	MT disorg.+IDA	Unknown	No	*CCDC40*	Chr17:78055687-78055691	Homoz	Frameshift	NM_017950.3	NP_060420.2	c.1819_1823delinsT	p.Leu607Trpfs*33	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G044	Not done	Other	Unk	*CCDC40*	Chr17:78032754	Homoz	Frameshift	NM_017950.3	NP_060420.2	c.1415delG	p.Arg472fs3*	5, 7	Absent	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G049	ODA	European	No	*CCDC40*	Chr17:78013765	Homoz	Frameshift	NM_017950.3	NP_060420.2	c.248delC	p.Ala83Valfs*84	5, 8	0.0004794	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G069	IDA	European	No	*CCDC40*	Chr17:78022417	Comp Het	Nonsense	NM_017950.3	NP_060420.2	c.712G>T	p.Glu238*	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP2
					Chr17:78023861		Splice acceptor	NM_017950.3	NP_060420.2	c.940-2A>G	p.?	7	0.00001661	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP2
PCD-G070	Not done	Other	No	*CCDC40*	Chr17:78013764-78013765	Comp Het	Frameshift	NM_017950.3	NP_060420.2	c.248delC	p.Ala83Valfs*84	5, 8	0.0004794	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP2
					Chr17:78061404		Splice acceptor	NM_017950.3	NP_060420.2	c.2450-2A>G	p.?	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP2
PCD-G086	MT disorg.+IDA	Arab	Yes	*CCDC40*	Chr17:78011940	Homoz	Synomynous creating exonic EES	NM_017950.3	NP_060420.2	c.48A>G	p.?	NR	Absent	Absent	Absent	Both parents carrier	3			PM2	PP1, PP4
PCD-G088	Not done	Arab	Yes	*CCDC40*	Chr17:78013904	Homoz	Nonsense	NM_017950.3	NP_060420.2	c.387C>G	p.Tyr129*	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G091	Not done	Arab	Yes	*CCDC40*	Chr17. Deletion of exon 11 & 12	Homoz	CNV	N/A	N/A	N/A	N/A	NR	N/A	N/A	N/A	No parental DNA	5	PVS1		PM2	PP2
PCD-G096	MT disorg.	European	No	*CCDC40*	Chr17:78063675	Comp Het	Frameshift	NM_017950.3	NP_060420.2	c.2824_2825insCTGT	p.Arg942Thrfs*57	NR	Absent	Absent	Absent	Fa carrier	5	PVS1		PM2	PP1, PP4
					Chr17:78069149		Nonsense	NM_017950.3	NP_060420.2	c.2920C>T	p.Gln974*	NR	Absent	Absent	Absent	Mo carrier	5	PVS1		PM2	PP1, PP4
PCD-G111	Not done	Arab	No	*CCDC40*	Chr17:78063675	Homoz	Frameshift	NM_017950.3	NP_060420.2	c.2824_2825insCTGT	p.Arg942Thrfs*57	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G031	MT disorg.	Other	Unk	*CCDC65*	Chr12:49312106	Homoz	Nonsense	NM_033124.4	NP_149115.2	c.658G>T	p.Glu220*	NR	8.243E-06	Absent	Absent	2 affected siblings, Mo carrier, no Fa DNA	5	PVS1		PM2	PP1, PP4
PCD-G019	Normal	Unknown	Yes	*CCDC103*	Chr17:42979917	Homoz	Missense	NM_001258395.1	NP_001245324.1	c.461A>C	p.His154Pro	11, 12, 13	0.001261	2 / 0.00101	6/0.0039	No parental DNA	5		PS3	PM1, PM2	PP2,PP3
PCD-G037	Normal	South-Asian	No	*CCDC103*	Chr17:42979917	Homoz	Missense	NM_001258395.1	NP_001245324.1	c.461A>C	p.His154Pro	11, 12	0.001261	2 / 0.00101	6/0.0039	No parental DNA	5		PS3	PM1, PM2	PP2,PP3
PCD-G057	IDA	European		*CCDC103*	Chr17:42979917	Homoz	Missense	NM_001258395.1	NP_001245324.1	c.461A>C	p.His154Pro	11, 12, 13	0.001261	2 / 0.00101	6/0.0039	No parental DNA	5		PS3	PM1, PM2	PP2,PP3
PCD-G068	IDA	South-Asian	Unk	*CCDC103*	Chr17:42979917	Homoz	Missense	NM_001258395.1	NP_001245324.1	c.461A>C	p.His154Pro	11, 12, 13	0.001261	2 / 0.00101	6/0.0039	Mo carrier, no Fa DNA	5		PS3	PM1, PM2	PP2,PP3
PCD-G078	IDA+ODA	South-Asian	Yes	*CCDC103*	Chr17:42979839-42979840	Homoz	Frameshift	NM_001258395.1	NP_001245324.1	c.383dupG	p.Pro129Serfs*25	25	Absent	Absent	1/0.0006	No parental DNA	5	PVS1	PS3	PM2	PP1, PP4
PCD-G082	IDA+ODA	Arab	Yes	*CCDC103*	Chr17:42978470	Homoz	Missense	NM_001258395.1	NP_001245324.1	c.104G>C	p.Arg35Pro	NR	8.241E-06	Absent	Absent	Both parents carrier	5		PS3	PM1, PM2	PP2-4
PCD-G116	IDA+ODA	South-Asian	Yes	*CCDC103*	Chr17:42979839-42979840	Homoz	Frameshift	NM_001258395.1	NP_001245324.1	c.383dupG	p.Pro129Serfs*25	25	Absent	Absent	1/0.0006	No parental DNA	5	PVS1	PS3	PM2	PP2
PCD-G012	Not done	South-Asian	Yes	*CCDC114*	Chr19:48805987	Homoz	Nonsense	NM_144577.3	NP_653178.3	c.1093C>T	p.Gln365*	NR	Absent	Absent	Absent	Fa carrier, no Mo DNA	4	PVS1		PM2	
PCD-G085	ODA	Arab	Yes	*CCDC151*	Chr19:11537077	Homoz	Nonsense	NM_145045.4	NP_659482.3	c.850C>T	p.Gln284*	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G080	IDA	South-Asian	Yes	*DRC1/ CCDC164*	Chr2. Deletion of exons 11, 12 & 13	Homoz	CNV	N/A	N/A	N/A	N/A	NR	N/A	N/A	N/A	No parental DNA	5	PVS1		PM2	PP4
PCD-G022	Lack of cilia	European	No	*CCNO*	Chr5:54528218	Homoz	Frameshift	NM_021147.3	NP_066970.3	c.538dupC	p.Val180Glyfs*55	NR	0.0000249	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G098	Lack of cilia	European	No	*CCNO*	Chr5:54529084	Homoz	Frameshift	NM_021147.3	NP_066970.3	c.263_267dup	p.Val90Serfs*6	26	Absent	Absent	Absent	Both parents carrier	5	PVS1	PS3	PM2	PP4

PCD-G075	IDA+ODA	Other	No	*DNAAF1*	Deletion of exon 1, 2 & 3	Homoz	CNV	N/A	N/A	N/A	N/A	NR	N/A	N/A	N/A	No parental DNA	5	PVS1		PM2	PP4
PCD-G001	IDA+ODA	South-Asian	No	*DNAAF3*	Chr19:55676764	Comp Het	Frameshift	NM_001256715.1	NP_001243644.1	c.296delT	p.Glu99Glyfs*88	NR	Absent	Absent	Absent	Mo carrier	5	PVS1		PM2	PP1, PP4
					Chr19:55673064-55673065		Inframe delins	NM_001256715.1	NP_001243644.1	c.609_610delinsTGGGA	p.Ala204delinsGlyThr	NR	Absent	Absent	Absent	Fa carrier	5	PVS1		PM2	PP1, PP4
PCD-G009	IDA+ODA	South-Asian	No	*DNAAF3*	Chr19:55677290-55677292	Homoz	Frameshift	NM_001256715.1	NP_001243644.1	c.162_164delinsG	p.Val55Glyfs*28	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G039	IDA+ODA	South-Asian	No	*DNAAF3*	Chr19: 55673053	Homoz	Frameshift	NM_001256715.1	NP_001243644.1	c.621dupT	p.Val208Cysfs*12	19	0.00008331	Absent	Absent	Fa carrier, no Mo DNA	5	PVS1	PS3	PM2	PP4
PCD-G047	IDA+ODA	European	No	*DNAAF3*	Chr19:55677221	Homoz	Splice site	NM_001256715.1	NP_001243644.1	c.228+5G>C	p.?	NR	0.00002956	Absent	Absent	No parental DNA	3			PM2	PP3, PP4
PCD-G048	IDA+ODA	European	No	*DNAAF3*	Chr19:55672025-55672026	Comp Het	Frameshift	NM_001256715.1	NP_001243644.1	c.1030_1031delinsG	p.Pro344Glyfs*64	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr19:55670783		Nonsense	NM_001256715.1	NP_001243644.1	c.1273G>T	p.Gly425*	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G002	ODA	European	No	*DNAH5*	Chr5:13701398	Comp Het	Nonsense	NM_001369.2	NP_001360.1	c.13486C>T	p.Arg4496*	1, 2	0.00005803	Absent	Absent	Mo carrier, no Fa DNA	5	PVS1	PS3	PM2	PP4
					Chr5:13701426		Frameshift	NM_001369.2	NP_001360.1	c.13458_13459insT	p.Asn4487fs*1	1, 3	0.00005783	Absent	Absent	Mo normal, no Fa DNA	5	PVS1	PS3	PM2	PP4
PCD-G013	Normal	European	No	*DNAH5*	Chr5:13708395	Comp Het	Missense	NM_001369.2	NP_001360.1	c.13175T>G	p.Phe4392Cys	NR	0.000486	Absent	Absent	No parental DNA	3			PM2	PP2,PP3
					Chr5:13870977		Missense	NM_001369.2	NP_001360.1	c.3733C>T	p.Arg1245Cys	NR	0.0002149	Absent	Absent	No parental DNA	3			PM2	PP2,PP3
PCD-G020	ODA	European	Unk	*DNAH5*	Chr5:13753599	Comp Het	Missense	NM_001369.2	NP_001360.1	c.10615C>T	p.Arg3539Cys	5, 14	Absent	Absent	Absent	No parental DNA	5		PS3	PM2, PM3	PP2-4
					Chr5:13753399		Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G023	IDA+ODA	European	No	*DNAH5*	Chr5:13817711-13817715	Comp Het	Frameshift	NM_001369.2	NP_001360.1	c.6930_6934delinsG	p.Asn2310Lysfs*15	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr5:13701426		Frameshift	NM_001369.2	NP_001360.1	c.13458_13459insT	p.Asn4487fs*1	1, 3	0.00005783	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G026	ODA	European		*DNAH5*	Chr5:13829802	Homoz	Nonsense	NM_001369.2	NP_001360.1	c.6261T>G	p.Tyr2087*	NR	Absent	Absent	Absent	2 affected siblings, no parents' DNA	5	PVS1		PM2	PP1, PP4
PCD-G028	Inconclusive	European	No	*DNAH5*	Chr5:13839639	Homoz	Splice acceptor	NM_001369.2	NP_001360.1	c.5710-2A>G	p.Cys1904-Lys1909del	14	0.00002482	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	
PCD-G029	ODA	European	No	*DNAH5*	Chr5:13737356	Comp Het	Splice site	NM_001369.2	NP_001360.1	c.11455+5G>A	p.?	NR	Absent	Absent	Absent	Mo carrier, no Fa DNA	4			PM2, PM3	PP3,PP4
					Chr5:13829802		Nonsense	NM_001369.2	NP_001360.1	c.6261T>G	p.Tyr2087*	NR	Absent	Absent	Absent	Mo normal, no Fa DNA	5	PVS1		PM2	PP4
PCD-G040	ODA	European	No	*DNAH5*	Chr5:13753399	Comp Het	Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
					Chr5:13701425-13701426		Frameshift	NM_001369.2	NP_001360.1	c.13458_13459insT	p.Asn4487fs*1	1, 3	0.00005783	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G043	ODA	European	No	*DNAH5*	Chr5:13753613	Comp Het	Missense	NM_001369.2	NP_001360.1	c.10601T>C	p.Phe3534Ser	NR	Absent	Absent	Absent	No parental DNA	4			PM2, PM3	PP2-4
					Chr5:13701425-13701426		Frameshift	NM_001369.2	NP_001360.1	c.13458_13459insT	p.Asn4487fs*1	1, 3	0.00005783	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G045	Not done	Other		*DNAH5*	Chr5:13886106	Homoz	Nonsense	NM_001369.2	NP_001360.1	c.2710G>T	p.Glu904*	NR	Absent	Absent	Absent	No parental DNA	4	PVS1		PM2	
PCD-G050	ODA	European	No	*DNAH5*	Chr5:13928248	Comp Het	Nonsense	NM_001369.2	NP_001360.1	c.232C>T	p.Arg78*	1, 2	Absent	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
					Chr5:13753399		Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G051	ODA	European	No	*DNAH5*	Chr5:13830872-13830873	Comp Het	Frameshift	NM_001369.2	NP_001360.1	c.5890_5894dup	p.Leu1966Serfs*9	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr5:13820505		Missense	NM_001369.2	NP_001360.1	c.6791G>A	p.Ser2264Asn	1	8.244E-06	Absent	Absent	No parental DNA	5		PS3	PM2, PM3	PP2-4
PCD-G052	ODA	European	No	*DNAH5*	Chr5:13708285	Comp Het	Nonsense	NM_001369.2	NP_001360.1	c.13285C>T	p.Arg4429*	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr5:13788830		Missense	NM_001369.2	NP_001360.1	c.8642C>G	p.Ala2881Gly	15	0.0000165	Absent	Absent	No parental DNA	5		PS3	PM2, PM3	PP2-4
PCD-G054	ODA	European	No	*DNAH5*	Chr5:13820533	Comp Het	Nonsense	NM_001369.2	NP_001360.1	c.6763C>T	p.Arg2255*	NR	0.00002472	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr5:13770983		Nonsense	NM_001369.2	NP_001360.1	c.9480T>A	p.Cys3160*	NR	8.242E-06	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G061	ODA	European	No	*DNAH5*	Chr5:13845040	Comp Het	Missense	NM_001369.2	NP_001360.1	c.5177T>C	p.Leu1726Pro	23	8.238E-06	Absent	Absent	Mo carrier, no Fa DNA	5		PS3	PM2	PP2-4
					Chr5:13902162		Missense	NM_001369.2	NP_001360.1	c.1730G>C	p.Arg577Thr	1	0.00003591	Absent	Absent	Mo normal, no Fa DNA	5		PS3	PM2	PP2-4
PCD-G063	Not done	European	No	*DNAH5*	Chr5:13792147	Comp Het	Nonsense	NM_001369.2	NP_001360.1	c.8404C>T	p.Gln2802*	1, 2	Absent	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	
					Chr5:13830135		Missense	NM_001369.2	NP_001360.1	c.6249G>A	p.Met2083Ile	NR	8.293E-06	Absent	Absent	No parental DNA	4			PM2, PM3	PP2, PP3
PCD-G065	ODA	European	No	*DNAH5*	Chr5:13753399	Homoz	Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G066	ODA	South-Asian	Yes	*DNAH5*	Chr5:13841167	Homoz	Nonsense	NM_001369.2	NP_001360.1	c.5557A>T	p.Lys1853*	NR	8.243E-06	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G079	ODA	European	No	*DNAH5*	Chr5:13753399	Comp Het	Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
					Chr5:13758990		Nonsense	NM_001369.2	NP_001360.1	c.10384C>T	p.Gln3462*	15	0.00004118	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G089	Not done	Arab	Yes	*DNAH5*	Chr5:13792231	Homoz	Missense	NM_001369.2	NP_001360.1	c.8320T>C	p.Trp2774Arg	NR	Absent	Absent	Absent	Both parents carrier	3			PM2	PP1-3
PCD-G092	Not done	Arab	Yes	*DNAH5*	Chr5:13737558	Comp Het	Frameshift	NM_001369.2	NP_001360.1	c.11258delT	p.Asn3753Thrfs*5	NR	Absent	Absent	Absent	Mo carrier	5	PVS1		PM2	PP1
					Chr5:13885116-13885117		Frameshift	NM_001369.2	NP_001360.1	c.2964_2965delGA	p.Thr990Asnfs*2	NR	Absent	Absent	Absent	Fa carrier	5	PVS1		PM2	PP1
PCD-G093	ODA	European	No	*DNAH5*	Chr5:13753399	Comp Het	Missense	NM_001369.2	NP_001360.1	c.10815T>G	p.Asp3605Glu	NR	Absent	Absent	Absent	Mo carrier, no Fa DNA	3			PM2	PP2,PP3
					Chr5:13845060		Synomynous creating exonic EES	NM_001369.2	NP_001360.1	c.5157C>T	p.?	NR	8.241E-06	Absent	Absent	Mo normal, no Fa DNA	3			PM2	PP3
PCD-G095	Normal	European	No	*DNAH5*	Chr5:13820514	Comp Het	Missense	NM_001369.2	NP_001360.1	c.6782T>G	p.Leu2261Arg	NR	Absent	Absent	Absent	2 affected siblings, Mo carrier, no Fa DNA	4			PM2, PM3	PP1-3
					Chr5:13914037		Nonsense	NM_001369.2	NP_001360.1	c.1351C>T	p.Gln451*	NR	Absent	Absent	Absent	2 affected siblings, Mo normal, no Fa DNA	5	PVS1		PM2	PP1-3
PCD-G108	ODA	European	No	*DNAH5*	Chr5:13762883	Comp Het	Missense	NM_001369.2	NP_001360.1	c.10229C>T	p.Thr3410Met	NR	0.00004118	Absent	Absent	Fa normal, no Mo DNA	4			PM2, PM3	PP2-4
					Chr5:13842004		Nonsense	NM_001369.2	NP_001360.1	c.5281C>T	p.Arg1761*	NR	0.00004119	Absent	Absent	Fa carrier, no Mo DNA	5	PVS1		PM2	PP4
PCD-G117	IDA+ODA	European	Unk	*DNAH5*	Chr5:13701426	*One hit*	Frameshift	NM_001369.2	NP_001360.1	c.13458_13459insT	p.Asn4487fs*1	1, 3	0.00005783	Absent	Absent	ND	NA	PVS1	PS3	PM2	PP4
PCD-G126	IDA+ODA	Unknown	Unk	*DNAH5*	Chr5:13753399	*One hit*	Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	ND	NA	PVS1	PS3	PM2	PP4
PCD-G128	ODA	European	No	*DNAH5*	Chr5:13753399	*One hit*	Frameshift	NM_001369.2	NP_001360.1	c.10815delT	p.Pro3606Hisfs*22	15	0.0001483	Absent	Absent	ND	NA	PVS1	PS3	PM2	PP4
PCD-G025	Normal	Unknown	Unk	*DNAH11*	Chr7:21723534	Homoz	Nonsense	NM_001277115.1	NP_001264044.1	c.5593C>T	p.Arg1865*	NR	0.00001657	Absent	1/0.0006	No parental DNA	5	PVS1		PM2	PP4
PCD-G032	Normal	European	No	*DNAH11*	Chr7:21675540	Comp Het	Nonsense	NM_001277115.1	NP_001264044.1	c.4552C>T	p.Gln1518*	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr7:21726874		Splice donor	NM_001277115.1	NP_001264044.1	c.5778+1G>A	p.Val1821Thrfs*7	17	0.00001193	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G036	Normal	European	No	*DNAH11*	Chr7:21789974	Comp Het	Nonsense	NM_001277115.1	NP_001264044.1	c.8932C>T	p.Gln2978*	NR	0.00001624	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4

					Chr7:21599378-21599383		Frameshift	NM_001277115.1	NP_001264044.1	c.853_857delinsG	p.Arg285Glufs*22	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G041	Inconclusive	South-Asian	Unk	*DNAH11*	Chr7:21934608	Comp Het	Missense	NM_001277115.1	NP_001264044.1	c.13040T>C	p.Leu4347Pro	NR	Absent	Absent	Absent	No parental DNA	4			PM2, PM3	PP2-4
					Chr7. Deletion of exons 68-75		CNV	N/A	N/A	N/A	N/A	NR	N/A	N/A	N/A	No parental DNA	5	PVS1		PM2	PP4
PCD-G072	Normal	European	No	*DNAH11*	Chr7:21727067	Comp Het	Missense	NM_001277115.1	NP_001264044.1	c.5846G>A	p.Arg1949Gln	NR	Absent	Absent	Absent	No parental DNA	4			PM2, PM3	PP2-4
					Chr7:21940699		Frameshift	NM_001277115.1	NP_001264044.1	c.13380_13383dup	p.Ala4462Leufs*22	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G073	Normal	European	No	*DNAH11*	Chr7:21824139-21824142	Comp Het	Frameshift	NM_001277115.1	NP_001264044.1	c.9581_9582delCT	p.Leu3194Glnfs*10	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr7:21658796		Nonsense	NM_001277115.1	NP_001264044.1	c.4333C>T	p.Arg1445*	NR	0.00003391	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G074	Normal	European	No	*DNAH11*	Chr7:21775289	Comp Het	Missense	NM_001277115.1	NP_001264044.1	c.7472G>C	p.Arg2491Pro	NR	0.000149	Absent	Absent	No parental DNA	5			PM2	PP2-4
					Chr7:21747335		Nonsense	NM_001277115.1	NP_001264044.1	c.6565C>T	p.Arg2189*	NR	0.00002484	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G100	Normal	Arab	Yes	*DNAH11*	Chr7:21932181	Homoz	Nonsense	NM_001277115.1	NP_001264044.1	c.12646G>T	p.Glu4216*	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G101	Normal	Other	Unk	*DNAH11*	Chr7:21641013	Comp Het	Splice acceptor	NM_001277115.1	NP_001264044.1	c.3426-1G>A	p.?	NR	0.00003394	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr7:21631042-21631056		Frameshift	NM_001277115.1	NP_001264044.1	c.2514_2528delinsC	p.Gln838Hisfs*26	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G104	Normal	Arab	Yes	*DNAH11*	Chr7:21598487	Homoz	Missense	NM_001277115.1	NP_001264044.1	c.563T>C	p.Met188Thr	NR	0.0000127	Absent	Absent	2 cousins, both parents carrier	3			PM2	PP1-4
PCD-G107	Normal	European	No	*DNAH11*	Chr7:21659634	Comp Het	Nonsense	NM_001277115.1	NP_001264044.1	c.4438C>T	p.Arg1480*	NR	0.00000828	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr7:21940815-21940821		Frameshift	NM_001277115.1	NP_001264044.1	c.13494_13500del	p.Ser4498Argfs*15	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G112	Not done	Arab	Yes	*DNAH11*	Chr7:21940815-21940821	Homoz	Frameshift	NM_001277115.1	NP_001264044.1	c.13494_13500del	p.Ser4498Argfs*15	NR	Absent	Absent	Absent	2 cousins, both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G115	Normal	South-Asian	Unk	*DNAH11*	Chr7:21920318	Comp Het	Splice acceptor	NM_001277115.1	NP_001264044.1	c.12196-2A>G	p.?	NR	Absent	Absent	Absent	2 affected siblings, no parental DNA	5	PVS1		PM2	PP1, PP4
					Chr7:21742391		Nonsense	NM_001277115.1	NP_001264044.1	c.6244C>T	p.Arg2082*	27	0.00004139	Absent	Absent	2 affected siblings, no parental DNA	5	PVS1	PS3	PM2	PP1, PP4
PCD-G118	Normal	European	No	*DNAH11*	Chr7:21726760	*One hit*	Missense	NM_001277115.1	NP_001264044.1	c.5665G>T	p.Gly1889Trp	NR	Absent	Absent	Absent	ND	NA			PM2	PP4
PCD-G119	Normal	European	No	*DNAH11*	Chr7:21745115	*One hit*	Nonsense	NM_001277115.1	NP_001264044.1	c.6506C>A	p.Ser2169*	28	Absent	Absent	Absent	ND	NA	PVS1		PM2	PP4
PCD-G123	Inconclusive	European	No	*DNAH11*	Chr7:21747434	*One hit*	Nonsense	NM_001277115.1	NP_001264044.1	c.6664C>T	p.Arg2222*	NR	8.277E-06	Absent	Absent	ND	NA	PVS1		PM2	PP4
PCD-G010	ODA	European	No	*DNAI1*	Chr9:34459053	Homoz	Splice donor	NM_012144.3	NP_036276.1	c.48+2dupT	p.Ser17Valfs*12	9	0.0004624	Absent	Absent	Both parents carrier	5	PVS1	PS3	PM2	PP1, PP4
PCD-G014	IDA+ODA	European	Yes	*DNAI1*	Chr9:34514434	Homoz	Missense	NM_012144.3	NP_036276.1	c.1612G>A	p.Ala538Thr	9	0.0000742	Absent	Absent	2 affected siblings, no parents' DNA	5		PS3	PM2	PP1-4
PCD-G016	ODA	European	No	*DNAI1*	Chr9: 34459053	Homoz	Splice donor	NM_012144.3	NP_036276.1	c.48+2dupT	p.Ser17Valfs*12	9	0.0004624	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G122	ODA	European	Unk	*DNAI1*	Chr9: 34459053	*One hit*	Splice site	NM_001281428.1	NP_001268357.1	c.48+2dupT	p.Ser17Valfs*12	9, 29	0.0004624	Absent	Absent	ND	NA	PVS1	PS3	PM2	PP4
PCD-G077	Normal	Unknown	Unk	*DNAI2*	Chr17:72306303	Homoz	Splice donor	NM_023036.4	NP_075462.3	c.1494+2dupT	p.?	NR	0.00000864	Absent	Absent	No parental DNA	4	PVS1		PM2	
PCD-G105	ODA	European	No	*DNAI2*	Chr17:72297203	Homoz	Nonsense	NM_023036.4	NP_075462.3	c.883C>T	p.Arg295*	NR	0.00001647	Absent	Absent	Fa carrier, no Mo DNA	5	PVS1		PM2	PP4
PCD-G027	Lack of cilia	European	No	*DYX1C1*	Chr15. Deletion of exon 7	Homoz	CNV	N/A	N/A	N/A	N/A	16	N/A	N/A	N/A	Mo carrier, no Fa DNA	5	PVS1	PS3	PM2	
PCD-G042	IDA+ODA	European	No	*DYX1C1*	Chr15. Deletion of exon 7	Homoz	CNV	N/A	N/A	N/A	N/A	16	N/A	N/A	N/A	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G121	IDA+ODA	South-Asian	No	*HEATR2*	Chr7:794249	*One hit*	Missense	NM_017802.3	NP_060272.3	c.1048C>T	p.Arg350Trp	NR	0.00007419	Absent	Absent	ND	NA			PM2	PP2, PP3
PCD-G017	Normal	South-Asian	No	*HYDIN*	Chr16:70894739	Comp Het	Missense	NM_001270974.1	NP_001257903.1	c.11843C>G	p.Pro3948Arg	NR	Absent	Absent	Absent	Not Sanger confirmed	3			PM2	PP2-4
					Chr16:70913319		Missense	NM_001270974.1	NP_001257903.1	c.10438G>A	p.Val3480Met	NR	Absent	Absent	Absent	Not Sanger confirmed	3			PM2	PP2-4
PCD-G021	Normal	European	No	*HYDIN*	Chr16:70989412	Comp Het	Missense	NM_001270974.1	NP_001257903.1	c.6182A>G	p.Asn2061Ser	NR	Absent	Absent	Absent	Not Sanger confirmed	3			PM2	PP2-4
					Chr16:71101211		Missense	NM_001270974.1	NP_001257903.1	c.2057C>T	p.Ala686Val	NR	Absent	Absent	Absent	Not Sanger confirmed	3			PM2	PP2-4
PCD-G120	Normal	South-Asian	No	*HYDIN*	Chr16:70916766	*One hit*	Nonsense	NM_001270974.1	NP_001257903.1	c.10012G>T	p.Glu3338*	NR	Absent	Absent	Absent	ND	NA	PVS1		PM2	PP4
PCD-G124	Normal	European	No	*HYDIN*	Chr16:70917997	*One hit*	Missense	NM_001270974.1	NP_001257903.1	c.9805T>G	p.Tyr3269Asp	NR	Absent	Absent	Absent	ND	NA			PM2	PP2-4
PCD-G125	Normal	Unknown	Unk	*HYDIN*	Chr16:71171147	*One hit*	Missense	NM_001270974.1	NP_001257903.1	c.950G>A	p.Arg317Gln	NR	0.00001845	Absent	Absent	ND	NA			PM2	PP2-4
PCD-G127	Normal	South-Asian	No	*HYDIN*	Chr16:70972589	*One hit*	Missense	NM_001270974.1	NP_001257903.1	c.6923A>T	p.Asp2308Val	NR	0.00001655	Absent	Absent	ND	NA			PM2	PP2-4
PCD-G006	IDA+ODA	South-Asian	Yes	*LRRC6*	Chr8:133645009	Homoz	Frameshift	NM_012472.4	NP_036604.2	c.630delG	p.Trp210Cysfs*12	5	0.000206	Absent	10/0.0065	Both parents carrier	5	PVS1	PS3	PM2	PP1, PP4
PCD-G033	Not done	South-Asian	Yes	*LRRC6*	Chr8:133645009	Homoz	Frameshift	NM_012472.4	NP_036604.2	c.630delG	p.Trp210Cysfs*12	5	0.000206	Absent	10/0.0065	Fa carrier, no Mo DNA	5	PVS1	PS3	PM2	
PCD-G046	IDA+ODA	South-Asian	No	*LRRC6*	Chr8:133645009	Homoz	Frameshift	NM_012472.4	NP_036604.2	c.630delG	p.Trp210Cysfs*12	5	0.000206	Absent	10/0.0065	No parental DNA	5	PVS1	PS3	PM2	
PCD-G059	IDA+ODA	South-Asian	Yes	*LRRC6*	Chr8:133645009	Homoz	Frameshift	NM_012472.4	NP_036604.2	c.630delG	p.Trp210Cysfs*12	5	0.000206	Absent	10/0.0065	Mo carrier, no Fa DNA	5	PVS1	PS3	PM2	
PCD-G071	IDA+ODA	South-Asian	Unk	*LRRC6*	Chr8:133645009	Homoz	Frameshift	NM_012472.4	NP_036604.2	c.630delG	p.Trp210Cysfs*12	5	0.000206	Absent	10/0.0065	No parental DNA	5	PVS1	PS3	PM2	
PCD-G083	IDA+ODA	Arab	Yes	*LRRC6*	Chr8:133645203	Homoz	Missense	NM_012472.4	NP_036604.2	c.436G>C	p.Asp146His	10	0.0001977	Absent	Absent	Both parents carrier	5		PS3	PM2	PP1-4
PCD-G090	Not done	Arab	Yes	*LRRC6*	Chr8:133627283	Homoz	Splice donor	NM_012472.4	NP_036604.2	c.974+1G>A	p.?	NR	Absent	Absent	Absent	No parental DNA	4	PVS1		PM2	
PCD-G058	Lack of cilia	Other	Yes	*MCIDAS*	Chr5:54518828-54518829	Homoz	Frameshift	NM_001190787.1	NP_001177716.1	c.332_333delinsG	p.Ala111Glyfs*22	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G084	Lack of cilia	South-Asian	Unk	*MCIDAS*	Chr5:54516635	Homoz	Splice acceptor	NM_001190787.1	NP_001177716.1	c.718-1G>A	p.?	NR	Absent	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
PCD-G035	Normal	Unknown	No	*OFD1*	ChrX:13785391-13785393	Hemizyg	Frameshift	NM_003611.2	NP_003602.1	c.2745_2747delinsC	p.Tyr916Serfs*7	NR	Absent	Absent	N/A	No parental DNA	4	PVS1		PM2	
PCD-G034	ODA	South-Asian	No	*PIH1D3*	ChrX:106462133	Hemizyg	Nonsense	NM_001169154.1	NP_001162625.1	c.266G>A	p.Trp89*	18	Absent	Absent	N/A	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G024	Lack of cilia	European	No	*RPGR*	ChrX:38170000	Hemizyg	Nonsense	NM_001034853.1	NP_001030025.1	c.646G>T	p.Glu216*	NR	Absent	Absent	N/A	2 affected siblings, no parents' DNA	5	PVS1		PM2	PP1
PCD-G015	Normal	European	No	*RSPH1*	Chr21:43906573	Homoz	Splice acceptor	NM_080860.3	NP_543136.1	c.275-2A>C	p.Gly92Alafs*10	10	0.0003625	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G062	CP	European	No	*RSPH1*	Chr21:43906573	Homoz	Splice acceptor	NM_080860.3	NP_543136.1	c.275-2A>C	p.Gly92Alafs*10	10	0.0003625	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G076	CP	European	Unk	*RSPH1*	Chr21:43906573	Comp Het	Splice acceptor	NM_080860.3	NP_543136.1	c.275-2A>C	p.Gly92Alafs*10	10	0.0003625	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
					Chr21:43906565		Nonsense	NM_080860.3	NP_543136.1	c.281G>A	p.Trp94*	24	Absent	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G097	MT disorg.	European	No	*RSPH1*	Chr21:43906573	Homoz	Splice acceptor	NM_080860.3	NP_543136.1	c.275-2A>C	p.Gly92Alafs*10	10	0.0003625	Absent	Absent	Fa carrier, no Mo DNA	5	PVS1	PS3	PM2	PP4
PCD-G004	CP	Other	Yes	*RSPH4A*	Chr6:116938246	Homoz	Nonsense	NM_001010892.2	NP_001010892.1	c.460C>T	p.Gln154*	4	0.00001647	Absent	3/0.00195	No parental DNA	5	PVS1	PS3	PM2	PP4

PCD-G038	CP	European	No	*RSPH4A*	Chr6:116953417	Homoz	Frameshift	NM_001010892.2	NP_001010892.1	c.1962_1966delinsC	p.Asp655Ilefs*83	NR	Absent	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1, PP4
PCD-G055	CP	European	No	*RSPH4A*	Chr6:116949221	Comp Het	Nonsense	NM_001010892.2	NP_001010892.1	c.1351C>T	p.Gln451*	NR	0.00004121	Absent	Absent	No parental DNA	5	PVS1		PM2	PP4
					Chr6:116937902		Nonsense	NM_001010892.2	NP_001010892.1	c.116C>A	p.Ser39*	21	0.00001657	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G064	CP	Arab	Yes	*RSPH4A*	Chr6:116949326	Homoz	Missense	NM_001010892.2	NP_001010892.1	c.1456G>C	p.Ala486Pro	NR	Absent	Absent	Absent	No parental DNA	3			PM2	PP2-4
PCD-G003	CP	Arab	Yes	*RSPH9*	Chr6:43638659-43638661	Homoz	Inframe del	NM_152732.4	NP_689945.2	c.801_803delGAA	p.Lys268del	4	0.00005765	3 / 0.00151	Absent	No parental DNA	5	PVS1	PS3	PM2, PM4	PP4
PCD-G030	MT disorg.	Arab	Unk	*RSPH9*	Chr6:43638659-43638661	Homoz	Inframe del	NM_152732.4	NP_689945.2	c.801_803delGAA	p.Lys268del	4	0.00005765	3 / 0.00151	Absent	No parental DNA	5	PVS1	PS3	PM2, PM4	PP4
PCD-G109	Not done	Arab	Yes	*RSPH9*	Chr6:43638611	Homoz	Frameshift	NM_152732.4	NP_689945.2	c.760delG	p.Arg254Alafs*76	NR	Absent	Absent	Absent	Fa affected, Mo carrier	5	PVS1		PM2	PP1
PCD-G053	ODA	Other	Yes	*SPAG1*	Chr8:101226129	Homoz	Frameshift	NM_003114.4	NP_003105.2	c.1519dupA	p.Ile507Asnfs*5	20	0.00004118	Absent	Absent	No parental DNA	5	PVS1	PS3	PM2	PP4
PCD-G011	IDA+ODA	Unknown	Yes	*ZMYND10*	Chr3:50381271	Homoz	Missense	NM_015896.2	NP_056980.2	c.212T>C	p.Leu71Pro	NR	Absent	Absent	Absent	3 affected siblings, no parents' DNA	4			PM2	PP1-4
PCD-G094	IDA+ODA	European	No	*ZMYND10*	Chr3:50380737	Homoz	Splice donor	NM_015896.2	NP_056980.2	c.510+1delC	p.?	NR	Absent	Absent	Absent	2 affected siblings, both parents carrier	5	PVS1		PM2	PP1-4
PCD-G099	Inconclusive	Other	Yes	*ZMYND10*	Chr3:50382928	Homoz	Missense	NM_015896.2	NP_056980.2	c.83G>A	p.Gly28Asp	NR	Absent	Absent	Absent	No parental DNA	3			PM2	PP2,PP3
PCD-G114	Not done	Arab	Yes	*ZMYND10*	Chr3:50380758	Homoz	Nonsense	NM_015896.2	NP_056980.2	c.490C>T	p.Gln164*	NR	0.00001667	Absent	Absent	Both parents carrier	5	PVS1		PM2	PP1

TEM, transmission electron microscopy; ODA, IDA, outer and inner dynein arms; MT disorg, microtubular disorganisation; CP, central pair defect; Unk, unknown; comp het, compound heterozygous; Cons, consanguineous family. Variant databases and pathogenicity classification described in Methods. HGMD (Human Genome Mutation Database Professional) references:1[11], 2[12], 3[13], 4[14], 5[15], 6[16], 7[17], 8[18], 9[19], 10[20], 11[21], 12[22], 13[23], 14[24], 15[25], 16[26], 17[27], 18[28], 19[29], 20[30], 21[31], 22[32], 23[33], 24[34], 25[35], 26[36], 27[37], 28[38], 29[9]. Predicted pathogenicity classifications and evidences (PVS, PS, PM, PP) follow the American College of Medical Genetics and Genomics (ACMG) Standards and Guidelines[39]. Variants have been classed as proven pathogenic (class 5), likely pathogenic (class 4) or of unknown clinical significance (VUS, class 3). NA, indicates that a pathogenicity classification was not possible based on the available evidence.

Table S4. Clinical data of 27 patients without prior TEM analysis where PCD still clinically suspected

Study ID	History of NRDS	Wet Cough	Rhinitis	Glue Ear	Situs abnormality
PCD-G012	Yes	Yes	Yes	No	Yes
PCD-G033	N/A	N/A	N/A	N/A	N/A
PCD-G044	No	No	No	No	No
PCD-G045	N/A	Yes	Yes	Yes	Yes
PCD-G148	Yes	Yes	Yes	Yes	No
PCD-G063	N/A	N/A	N/A	N/A	Yes
PCD-G070	N/A	N/A	N/A	N/A	N/A
PCD-G150	Yes	Yes	Yes	yes	No
PCD-G087	Yes	Yes	Yes	Yes	Yes
PCD-G152	Yes	Yes	Yes	No	Yes
PCD-G153	N/A	Yes	Yes	No	Yes
PCD-G154	N/A	Yes	Yes	N/A	No
PCD-G138	No	Yes	Yes	No	Yes
PCD-G088	Yes	Yes	Yes	No	Yes
PCD-G139	Yes	Yes	Yes	No	Yes
PCD-G089	Yes	Yes	Yes	Yes	Yes
PCD-G090	Yes	Yes	Yes	No	Yes
PCD-G091	No	Yes	Yes	Yes	Yes
PCD-G092	Yes	Yes	Yes	Yes	Yes
PCD-G157	N/A	N/A	N/A	N/A	N/A
PCD-G109	Yes	Yes	Yes	Yes	No
PCD-G158	Yes	Yes	Yes	Yes	Yes
PCD-G110	Yes	Yes	Yes	No	Yes
PCD-G111	Yes	Yes	Yes	Yes	Yes
PCD-G112	Yes	Yes	Yes	Yes	Yes
PCD-G113	Yes	Yes	Yes	No	Yes
PCD-G114	N/A	N/A	N/A	N/A	N/A

NRDS; neonatal respiratory distress syndrome, N/A; data not available.

Table S5. Consanguinity among different ethnicities

Ethnicity	No. of families	Consanguinity		
		Yes	No	Unknown
European	74	1	66	7
South-Asian	35	12	14	9
Arab	29	25	3	1
Other	15	7	4	4
Unknown	8	2	2	4

Ethnicity details for 161 families enrolled in this study, recruited through the UK national PCD diagnostic and management service in addition to those recruited by clinical collaborators in Portugal, Palestine and Egypt. Arab families included families from Egypt, Palestine, Kuwait and Iraq. South Asian families are originally from Pakistan, India, Bangladesh, Nepal and Sri Lanka. Other families are from Turkey, Chile, Afro-Caribbean, Somalia, West Africa, South Africa, Hong Kong and mixed ethnicities.

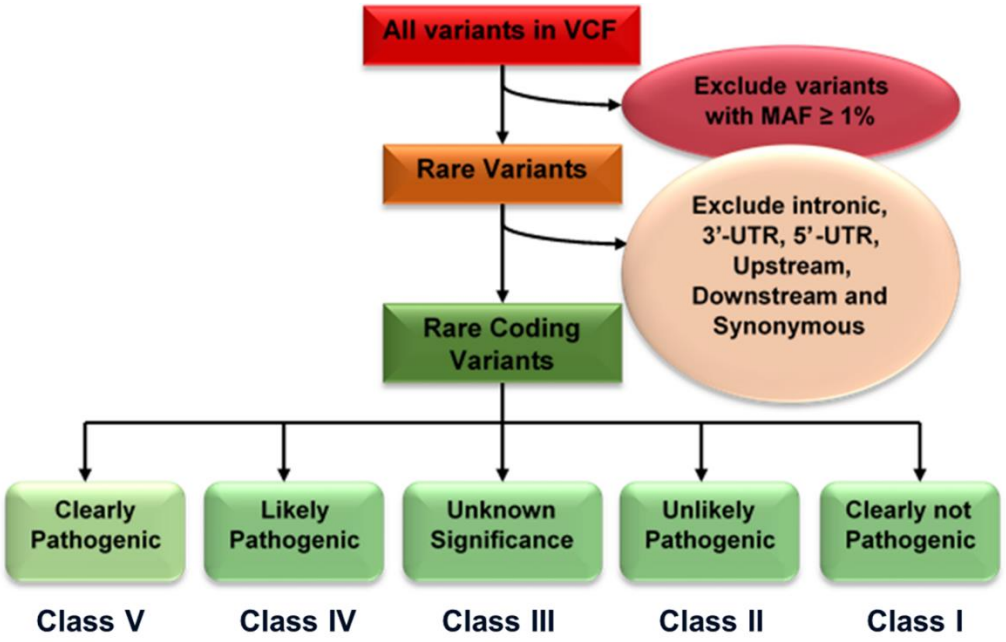


Figure S1. Variant filtration and prioritization workflow. All variants with MAF ≥1% were excluded followed by filtering out all variants called within intronic, 3'-UTR, 5'-UTR, upstream, downstream regions. All synonymous variants were firstly filtered out and re-visited in cases only where it was not possible to prioritize any other potentially pathogenic variants, in these cases the variants were re-assessed based on their potential to cause splicing defects.

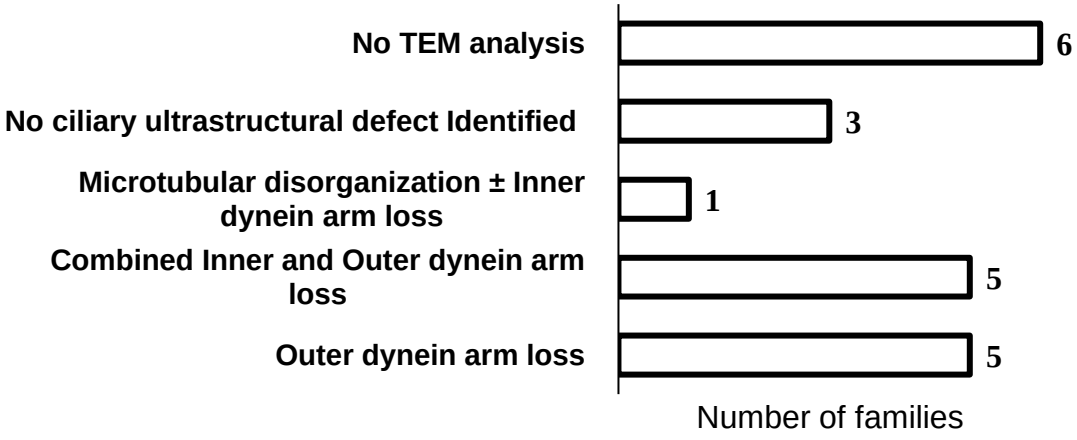
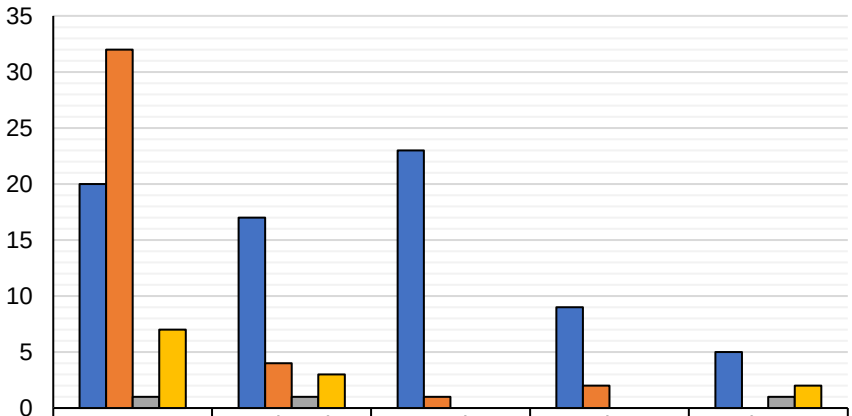


Figure S2. Transmission electron microscopy findings in 20 families with no genetic results. TEM analysis was not available for 6 of these families. In the other 14 families with TEM results, an ultrastructural defect was identified in 11 families, while in 3 families inconclusive TEM results were reported.



	European	South-Asian	Arab	Other	Unknown
Genotype Homozygous	20	17	23	9	5
Genotype Comp. Heterozygous	32	4	1	2	0
Genotype Hemizygous	1	1	0	0	1
Genotype One mutant allele	7	3	0	0	2

Figure S3. Genotype status of mutations in known PCD genes among different ethnicities. In Arab and South-Asian families, the majority of variants in known PCD genes were homozygous. One third of variants identified in European populations were also homozygous (20 out of 60 families).

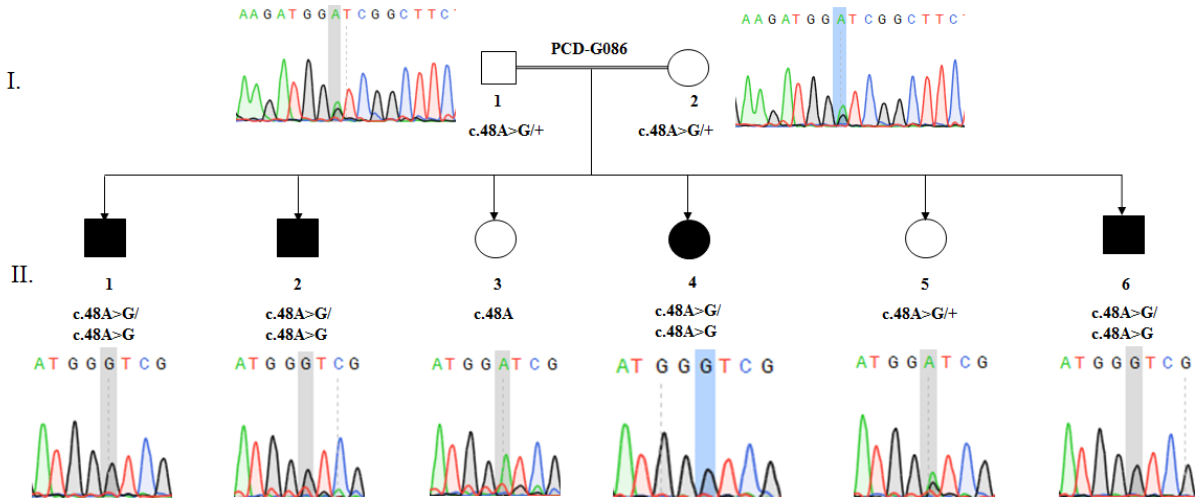


Figure S4. Familial segregation of a *CCDC40* synonymous variant in Arab family with microtubular disorganization and IDA loss

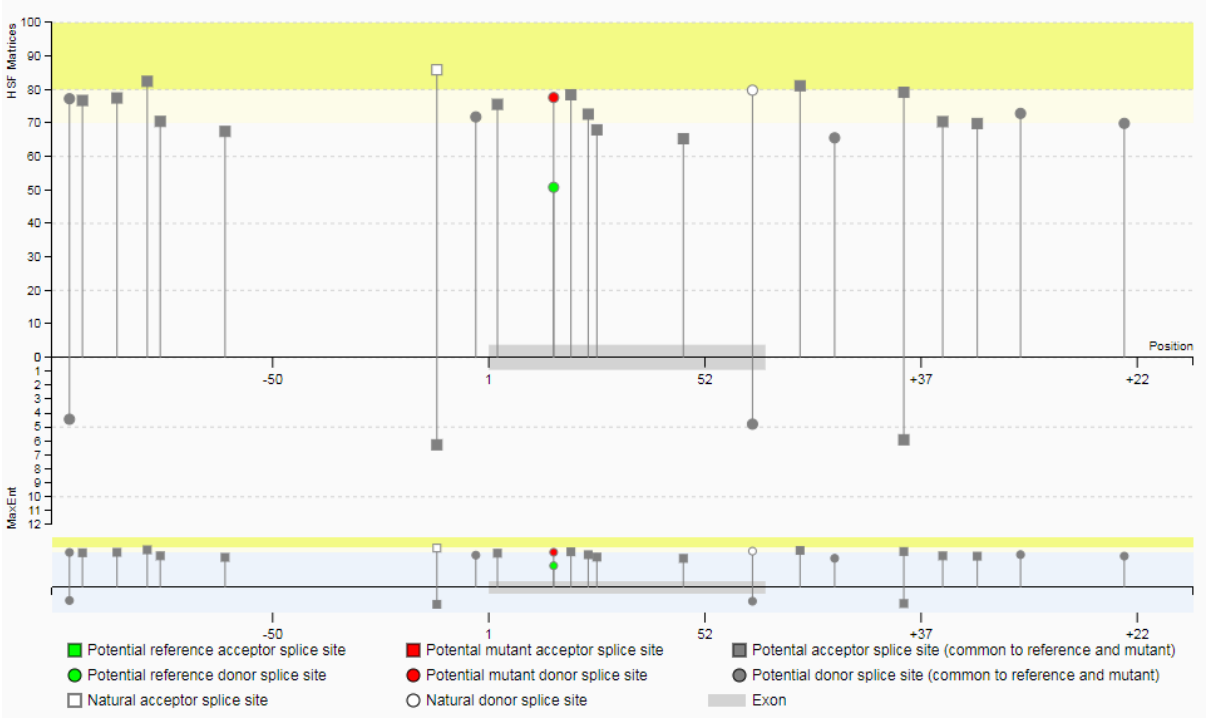


Figure S5. Graphical representation of the predicted exonic cryptic donor site associated with a c.48A>G synonymous variant in *CCDC40*. Analysis used the HSF Matrices algorithm in the Human Splicing Finder 3.0 tool which identified the predicted mutant splice site (red circle).

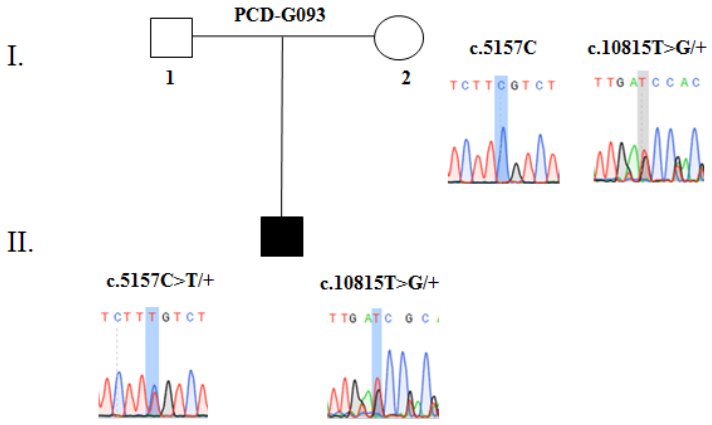


Figure S6. *DNAH5* compound heterozygous variants in a European family with ODA loss. One variant (c.5157C>T; p.Phe1719Phe) is a synonymous variant affecting splicing (**Figure S7**). The other variant (c.10815T>G; p.Asp3605Glu) is a missense variant. Paternal DNA was not available to confirm carrier state of the synonymous variant.

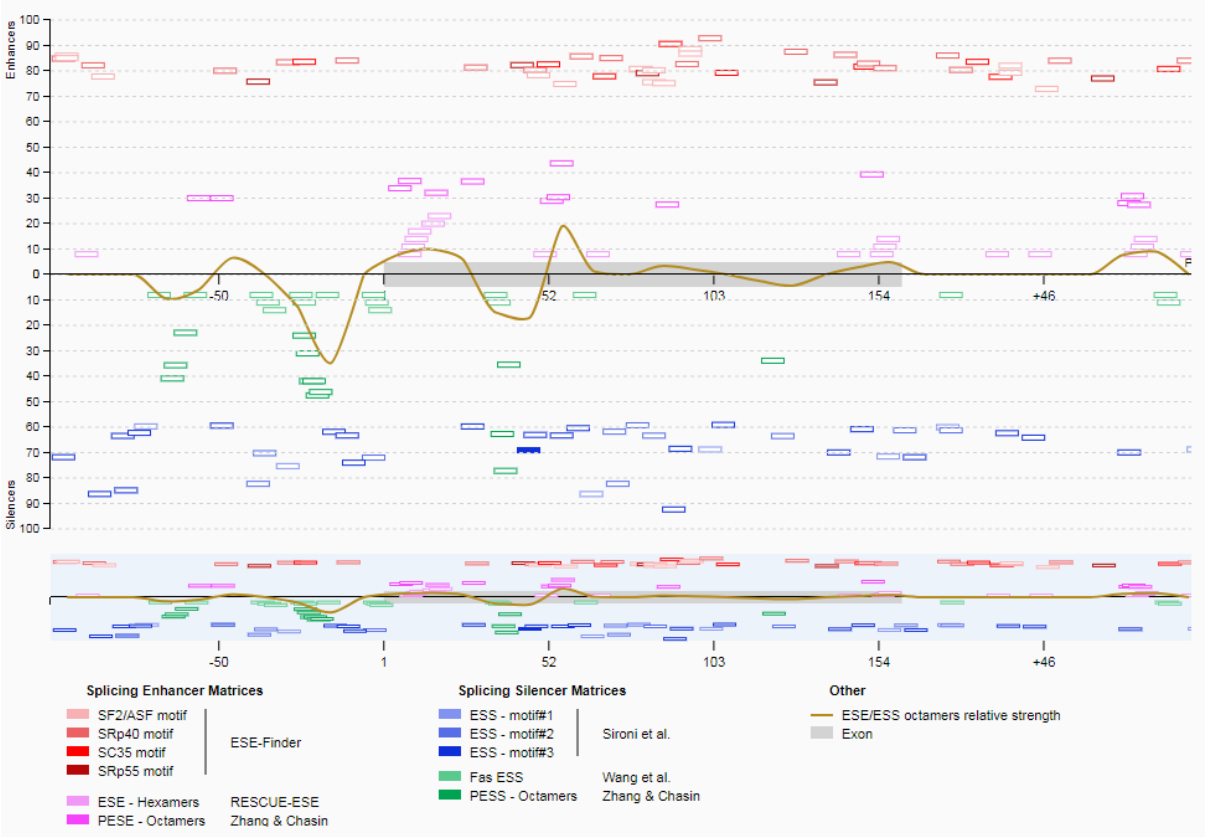


Figure S7. Graphical representation of the predicted creation of a new exonic splicing silencer due to c.5157C>T; p.Phe1719Phe synonymous variant in *DNAH5*. Variant found in the European family shown in **Figure S6**, effect predicted using Human Splicing finder 3.0 tool.

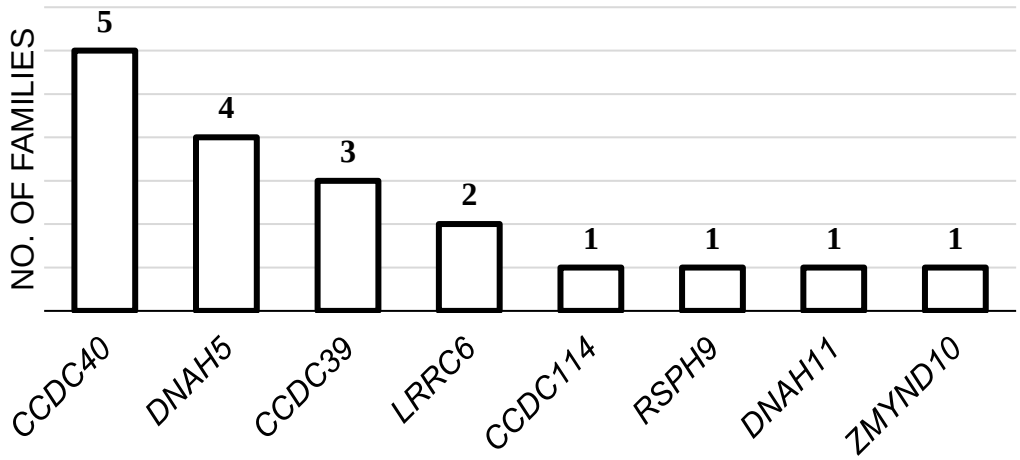


Figure S8. Mutant PCD genes in patients without TEM analysis. Of 161 families with PCD confirmed or highly suggested through diagnostic workup in this study, 17% (27 families) did not have available TEM results (not available or insufficient sample obtained from nasal brushing). Biallelic mutations in known PCD genes were identified in 18 of these families (67%).

Web Resources

CILDB	http://cildb.cgm.cnrs-gif.fr/
dbSNP build 141	https://www.ncbi.nlm.nih.gov/projects/SNP/
ExAC	http://exac.broadinstitute.org/
Exome Variant Server	http://evs.gs.washington.edu/EVS/
1000Genomes	http://1000genomes.org/
NCBI Primer-BLAST tool	https://www.ncbi.nlm.nih.gov/tools/primer-blast/
Human Splicing Finder	http://www.umd.be/HSF3/
SIFT	http://sift.jcvi.org/
Polyphen-2	http://genetics.bwh.harvard.edu/pph2/
Mutation Taster	http://www.mutationtaster.org/
CADD	http://cadd.gs.washington.edu/
MAPP	http://mendel.stanford.edu/sidowlab/downloads/MAPP/index.html
PhastCons	http://compgen.cshl.edu/phast/
PhyloP	http://compgen.bscb.cornell.edu/phast/help-pages/phyloP.txt
Phytozome	https://phytozome.jgi.doe.gov
BLAST/BLOSUM62	https://blast.ncbi.nlm.nih.gov/Blast.cgi
OMIM	http://www.omim.org/
RefSeq	http://www.ncbi.nlm.nih.gov/RefSeq
Ensembl Genome Browser	http://www.ensembl.org/index.html
NCBI	https://www.ncbi.nlm.nih.gov/

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