

#	Gene (Alias)	Chromosomal Location	Structure(s) Affected	Ciliary Ultrastructure by Transmission Electron Microscopy
1	DNAH5	5p15.2	Outer dynein arm	Outer dynein arm defects
2	DNAI1	9p13.3	Outer dynein arm	Outer dynein arm defects
3	DNAI2	17q25.1	Outer dynein arm	Outer dynein arm defects
4	DNAL1	14q24.3	Outer dynein arm	Outer dynein arm defects
5	ODAD1 (CCDC114)	19q13.33	Outer dynein arm	Outer dynein arm defects
6	ODAD2 (ARMC4)	10p12.1	Outer dynein arm	Outer dynein arm defects
7	ODAD3 (CCDC151)	19p13.2	Outer dynein arm	Outer dynein arm defects
8	ODAD4 (TTC25)	17q21.2	Outer dynein arm	Outer dynein arm defects
9	NME8 (TXNDC3)	7p14.1	Outer dynein arm	Partial outer dynein arm defects
10	ODAD5 (CLXN)	8q11.21	Outer dynein arm	Partial outer dynein arm defects
11	CCDC103 (DNAAF17)	17q21.31	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
12	CFAP298 (C21orf59, DNAAF16)	21q22.11	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
13	CFAP300 (C11orf70, DNAAF17)	11q22.1	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
14	DNAAF1 (LRRC50)	16q24.1	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
15	DNAAF11 (LRRC6)	8q24.22	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
16	DNAAF2 (KTU)	14q21.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
17	DNAAF3	19q13.42	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
18	DNAAF4 (DYG1C1)	15q21.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
19	DNAAF5 (HEATR2)	7p22.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
20	DNAAF6 (PIH1D3)	Xq22.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
21	SPAG1 (DNAAF13)	8q22.2	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
22	ZMYND10 (DNAAF7)	3p21.31	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
23	CCDC39	3q26.33	Inner dynein arm and Nexin links	Inner dynein arm defect and Microtubular disorganization
24	CCDC40	17q25.3	Inner dynein arm and Nexin links	Inner dynein arm defect and Microtubular disorganization
25	TTC12	11q23.2	Axonemal, Dynein arm assembly	Subtle Inner dynein arm defect
26	CCNO	5q11.2	Motile ciliogenesis	Sparse cilia
27	MCIDAS	5q11.2	Motile ciliogenesis	Sparse cilia
28	FOXJ1	17q25.1	Motile ciliogenesis	Sparse cilia or near normal
29	CCDC65 (DRC2)	12q13.12	Nexin Dynein Regulatory Complex	Near normal
30	DRC1 (CCDC164)	2p23.3	Nexin Dynein Regulatory Complex	Near normal
31	GAS8 (DRC4)	16q24.3	Nexin Dynein Regulatory Complex	Near normal
32	CFAP221 (PCDP1)	2q14.2	Central complex	Near normal
33	HYDIN	16q22.2	Central complex	Near normal
34	SPEF2	5p13.2	Central complex	Near normal
35	STK36	2q35	Central complex	Near normal
36	CFAP54	12q23.1	Central complex	Near normal
37	CFAP74	1p36.33	Central complex	Near normal
38	DNAJB13	11q13.4	Radial spoke	Near normal
39	NME5 (RSPH23)	5q31.2	Radial spoke	Near normal
40	RSPH1	21q22.3	Radial spoke	Near normal

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41	RSPH3	6q25.3	Radial spoke	Near normal
42	RSPH4A	6q22.1	Radial spoke	Near normal
43	RSPH9	6p21.1	Radial spoke	Near normal
44	DNAH11	7p15.3	Outer dynein arm	Near normal
45	DNAH9	17p12	Outer dynein arm	Near normal
46	DNAH1	3p21.1	Inner dynein arm	Unknown (data not available)
47	CFAP57 (WDR65)	1p34.2	Inner dynein arm assembly process	Near normal
48	GAS2L2	17q12	Basal body/basal foot/rootlet/actin filament	Near normal
49	OFD1	Xp22.2	Centrosome/basal body	Near normal
50	RPGR	Xp11.4	Transition zone of respiratory cilia	Near normal
51	LRRCS6 (DNAAF12)	11p15.5	Cargo for Intraflagellar transport	Near normal
52	NEK10	3p24.1	Protein kinase	Near normal
53	TP73	1p36.32	Transcription regulatory factor	Near normal
54	IFT74	9p21.2	Intraflagellar transport	Short and sparse cilia and Microtubular disorganization
55	TUBB4B	9q34.3	Microtubule	Sparse cilia with bulbous tip
56	KIAA0586 (TALPID3)	14q23.1	Centrosome	Short and sparse cilia with bulbous tip

For the current listing of laboratories offering clinical genetic testing for PCD, please check GTR: Genetic Testing Registry (<http://www.ncbi.nlm.nih.gov/gtr/>) and/or Gene Tests™ (<http://www.genetests.org>) Laboratory Directory.