

PCD Gene Listing February 2022

#	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	CCDC103 (DNAAF17)	17q21.31	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
11	CFAP298 (C21orf59, DNAAF16)	21q22.11	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
12	CFAP300 (C11orf70, DNAAF17)	11q22.1	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
13	DNAAF1 (LRRC50)	16q24.1	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
14	DNAAF11 (LRRC6)	8q24.22	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
15	DNAAF2 (KTU)	14q21.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
16	DNAAF3	19q13.42	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
17	DNAAF4 (DYX1C1)	15q21.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
18	DNAAF5 (HEATR2)	7p22.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
19	DNAAF6 (PIH1D3)	Xq22.3	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
20	SPAG1 (DNAAF13)	8q22.2	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
21	ZMYND10 (DNAAF7)	3p21.31	Axonemal, Dynein arm assembly	Outer and Inner dynein arm defects
22	CCDC39	3q26.33	Inner dynein arm and Nexin links	Inner dynein arm defect and Microtubular disorganization
23	CCDC40	17q25.3	Inner dynein arm and Nexin links	Inner dynein arm defect and Microtubular disorganization
24	TTC12	11q23.2	Axonemal, Dynein arm assembly	Inner dynein arm defect
25	CCNO	5q11.2	Motile ciliogenesis	Sparse cilia
26	MCIDAS	5q11.2	Motile ciliogenesis	Sparse cilia
27	FOXJ1	17q25.1	Motile ciliogenesis	Sparse cilia or near normal
28	CCDC65 (DRC2)	12q13.12	Nexin Dynein Regulatory Complex	Near normal
29	DRC1 (CCDC164)	2p23.3	Nexin Dynein Regulatory Complex	Near normal
30	GAS8 (DRC4)	16q24.3	Nexin Dynein Regulatory Complex	Near normal
31	CFAP221 (PCDP1)	2q14.2	Central complex	Near normal
32	HYDIN	16q22.2	Central complex	Near normal
33	SPEF2	5p13.2	Central complex	Near normal
34	STK36	2q35	Central complex	Near normal
35	DNAJB13	11q13.4	Radial spoke	Near normal
36	NME5 (RSPH23)	5q31.2	Radial spoke	Near normal
37	RSPH1	21q22.3	Radial spoke	Near normal
38	RSPH3	6q25.3	Radial spoke	Near normal

PCD Gene Listing February 2022

#	Gene (Alias)	Chromosomal Location	Structure(s) Affected	Ciliary Ultrastructure by Transmission Electron Microscopy
39	RSPH4A	6q22.1	Radial spoke	Near normal
40	RSPH9	6p21.1	Radial spoke	Near normal
41	DNAH11	7p15.3	Outer dynein arm	Near normal
42	DNAH8	6p21.2	Outer dynein arm	Unknown (data not available)
43	DNAH9	17p12	Outer dynein arm	Near normal
44	DNAH1	3p21.1	Inner dynein arm	Unknown (data not available)
45	CFAP57 (WDR65)	1p34.2	Inner dynein arm assembly process	Near normal
46	GAS2L2	17q12	Basal body/basal foot/rootlet/actin filament	Near normal
47	OFD1	Xp22.2	Centrosome/basal body	Near normal
48	RPGR	Xp11.4	Transition zone of respiratory cilia	Near normal
49	LRRC56 (DNAAF12)	11p15.5	Intraflagellar transport	Near normal
50	NEK10	3p24.1	Protein kinase	Near normal
51	TP73	1p36.32	Transcription regulatory factor	Near normal

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