Title: Primary Ciliary Dyskinesia GeneReview – Table 2
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Updated: December 2019

Table 2. Founder Variants by Ethnicity Reported in Individuals with Primary Ciliary Dyskinesia

Ethnicity (Specific Location)	Gene	DNA Nucleotide Change	Predicted Protein Change [1]	Reference Sequence
Amish &/or Mennonite [2]	*DNAAF5*	c.2384T>C	p.Leu795Pro	NM_017802.3
	DNAH5	c.10815delT	p.Pro3606HisfsTer23	NM_001369.2
	DNAH5	c.4348C>T	p.Gln1450Ter	NM_001369.2
	DNAI1	c.48+2dupT	--	NM_012144.3
Ashkenazi Jewish [3, 4]	*CFAP298*	c.735C>G	p.Tyr245Ter	NM_021254.3
	DNAI2	c.1304G>A	p.Trp435Ter	NM_023036.4
	CCDC65	c.877_878delAT	p.Ile293ProfsTer2	NM_033124.4
Asian [5]	*DRC1*	c.1-3952_540+1331del 27748-bp	--	NM_145038.4
Bedouin [6, 7]	*DNAL1*	c.449A>G	p.Asn150Ser	NM_031427.3
	RSPH9	c.804_806delGAA	p.Lys268del	NM_152732.4
Dutch (Volendam) [8]	*CCDC114*	c.742G>A	p.Ala248Thr	NM_144577.3
Faroe Islands [9]	*HYDIN*	c.922A>T	p.Lys308Ter	NM_001270974.1
Irish Traveler [10]	*CCNO*	c.258_262dupGGCC	p.Gln88ArgfsTer8	NM_021147.4
	RSPH4A	c.166dupC	p.Arg56ProfsTer11	NM_001010892.2
	DNAAF4	3549-bp large deletion	--	
Pakistani (United Kingdom)	*LRRC6*	c.630delG [11]	p.Trp210CysfsTer12	NM_012472.4
	RSPH4A	c.460C>T [7]	p.Gln154Ter	NM_001010892.2
	CCDC103	c.383dupG [12]	p.Pro129SerfsTer25	NM_213607.2
	CCDC103	c.461A>C [12, 13]	p.His154Pro	NM_213607.2
Puerto Rican [14]	*RSPH4A*	c.921+3_921+6delAAGT	--	NM_001010892.2
Polish [15]	*CFAP300*	c.198_200delTTTinsCC	p.Phe67ProfsTer10	NM_032930.2

1. As predicted by Mutalyzer name checker
2. Ferkol et al [2013]
3. Austin-Tse et al [2013]
4. Fedick et al [2014]
5. Hijikata et al [2019]
6. Mazor et al [2011]
7. Castleman et al [2009]
8. Onoufriadis [2013]
9. Olbrich et al 2012]
10. Casey et al [2015]
11. Zariwala et al [2013]
12. Panizzi et al [2012]
13. Shoemark et al [2018]
14. Daniels et al [2013]
15. Ziétkiewicz et al [2019]

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