Table SI. Primers used for pedigree and minigene analyses.

Primer ID	Sequence (5' to 3')
Pedigree analysis	
DNAH9-3743+1-F	GCACCTTGGGAATCTGACCA
DNAH9-3743+1-R	CCACGTTTCAACCAAAGGTCC
DNAH9-11176-F	TGGGCTGGAAAACTATCACATT
DNAH9-11176-R	TAAGTGGGAAATGGTGGGGG
Minigene plasmid construction[a]	
DNAH9-Exon18-F	tggccatggaggcccgaattCTTGGCCAACCTGGATGCG
DNAH9-Intron19-R	gggcaattcaaagatgcttcTAGATATATATGCATCTTTTT
DNAH9-Intron19-F	gaagcatctttgaattgcccTTCTTCCAATG
DNAH9-Exon20-R	gcggccgcggtacctcgagaCTGGGGTAGCTGTGCCCG
Minigene assay	
DNAH9-MG-test-F	GGCCAACCTGGATGCGTTTA
DNAH9-MG-test-R	CAGACCTCCTTCCTGCACTG

[a]Exon 18 + intron 18-19 + exon 19 + intron 19-20 + exon 20; lowercase letters represent the homologous arms, where the lowercase letters in the DNAH9-Exon18-F and DNAH9-Exon20-R primers indicate sequences complementary to the vector. Since the sequence of intron 19 of DNAH9 is too long, it was divided into two segments for cloning, and the sequences of the first 250 bp and the last 250 bp of intron 19 were retained, respectively. The underlined lowercase and uppercase letters in the DNAH9-Intron19-R primer represent the terminal sequence of the first DNAH9 cloning fragment, while the underlined lowercase and uppercase letters in the DNAH9-Intron19-F primer represent the beginning sequence of the second cloning fragment. *DNAH9*, dynein axonemal heavy chain 9; F, forward; R, reverse; MG, minigene.

Table SII. Frequency and pathogenicity analysis of dynein axonemal heavy chain 9 mutations.

	Frequency				**Pathogenicity score, prediction**			
Mutations	**1000 GEA**	**ESP6500**	**gnomAD**	**ExAC**	**PolyPhen-2**	**MutationTaster**	**SIFT**	**CADD**
c.11176C>T	0.0069	$7.7x10^{-5}$	0.0050	0.0055	1.000, probably damaging	1.0, disease causing	0.000, damaging	27.5, damaging
c.3743+1G>T	-	-	-	-	-	1.0, damaging	-	25.9, damaging

1000 GEA, 1000 Genomes (East Asian); ESP6500, Exome Sequencing Project 6500; gnomAD, The Genome Aggregation Database (East Asian); ExAC, Exome Aggregation Consortium (East Asian); SIFT, Sorting Intolerant from Tolerant; CADD, Combined Annotation Dependent Depletion.

Table SIII. Phenotypes of 24 patients with dynein axonemal heavy chain 9 gene mutations.

Individual	Phenotypes	Mutation sites	(Refs.)
P1	Asthenozoospermia	c.302dupT (p.Leu101fs*47) / c.6956A>G (p.Asp2319Gly)	(9)
P2	Asthenozoospermia	c.6294T>A (p.Phe2098Leu) / c.10571T>A (p.Leu3524Gln)	(9)
P3	Rhinosinusitis, situs inversus, CHD	c.12367G>A (p.Asp4123Asn) homo	(10)
P4	Rhinosinusitis, situs inversus	c.8708-2A>G (p.Glu2904Aspfs*53) / c.10193G>T (p.Arg3398Leu)	(10)
P5	Rhinosinusitis, situs inversus	c.8708-2A>G (p.Glu2904Aspfs*53) / c.10193G>T (p.Arg3398Leu)	(10)
P6	Rhinosinusitis, asthenozoospermia, situs inversus	c.5641A>G (p.Lys1881Glu) homo / c.8894G>A (p.Arg2965His) homo	(10)
P7	Levocardia, SV, MA, DORV, PS, liver/spleen inversion	c.5308A>T (p.K1770Ter) / c.5381T>C (p.I1794T)	(11)
P8	Dextrocardia, SAV, ASD, TGA, PS	c.4703A>C (p.N1568T) / c.11009A>G (p.E3670G)	(11)
P9	Dextrocardia. PLSVC, TGA, VSD, DORV, PS	c.12844-1G>C (splice acceptor) / c.12080A>T (p.H4027L)	(11)
P10	Isolated levocardia, liver/spleen inversion, MA, ASD, SV, TGA, PS	c.302delT (p.F103SfsTer31) / c.5381T>C (p.I1794T)	(11)
P11	TGA, PS, no laterality defects	c.302dupT (p.L104Pfs*45) / c.11176C>T (p.R3726W)	(11)
P12	Laterality defects	c.12914_12919del (p.4305_4307delCCT GGG) / c.7682A>T (p.H2561L)	(11)
P13	VSD, PS	c.4394A>T (p.D1465V) / c.8683C>T (p.L2895F)	(11)
P14	Complete endocardial cushion defect, DORV	c.3918G>A (p.Trp1306ter) / c.7150G>A (p.Gly2384Arg)	(11)
P15	SIT	c.1970+4A>G / c.3354-1G>T	(12)
P16	SIT	c.8251C>T (p.Gln2751ter) homo	(12)
P17	SIT	c.1027dupT (p.Leu3376Phefs*57) homo	(12)
P18	Situs ambiguous with CHD (LAI, ACD, IVCI, APVC, PDA)	c.308dupT (p.Leu104Profs*45) / c.11666C>G (p.Ser3889ter)	(12)
P19	Heterotaxy	c.1997G>A (p.Trp666ter) / c.5020G>A (p.Gly1674Arg)	(12)
P20	CHD	81-kb deletion at chr17p12 (11,486,795-11,568,385) c.10975C>T (p.Q3659*)	(13)
P21	Recurrent cough, situs inversus	c.760_761delTT (p.Phe254Leufs*6) c.4331G>A	(14)
P22	Repeated episodes of productive cough	c.1298C>G (p.Ser433Cys) / c.5547_5550delTGAC (p.asp1850fs)	(15)
P23	Primary ciliary dyskinesia, jejunal atresia	c.7150G>A (p.Gly2384Arg) / c.11086C>T (p.His3696Tyr)	(16)
P24	Heterotaxy of abdominal organs, intrauterine fetal death	c.12775T>C (p.Cys4259Arg) / RSPH1, c.121G>A (p.G41R)	(17)

ACD, atrioventricular canal defect; APVC, anomalous pulmonary vein connection; ASD, atrioventricular septal defect; CHD, congenital heart disease; DORV, double outlet right ventricle; IVCI, interrupted vena cava inferior; LAI, left atrial isomerism; MA, mitral atresia; PDA, persistent ductus arteriosus; PLSVC, persistent left superior vena cava; PS, pulmonary stenosis; SAV, single atrium and ventricle; SIT, situs inversus totalis; SV, single ventricle; TGA, transposition of the great arteries; VSD, ventricular septal defect.