

Laterality Defects Other Than Situs Inversus Totalis in Primary Ciliary Dyskinesia

Insights Into Situs Ambiguus and Heterotaxy

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e-Appendix 1. IRB approvals from all participating sites

The University of North Carolina at Chapel Hill

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

Medical IRB Study # 05-2979 (GCRC#2486)

Committee Name: Office of Human Research Ethics

Washington University in St. Louis

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests – Protocol Version May 11

Medical IRB Study # 201011855

Committee Name: The Washington University in St. Louis Institutional Review Board

University of Colorado, Denver; Children’s Hospital Colorado

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

Medical IRB Study # 04-1106

Committee Name: COMIRB - Colorado Multiple Institutional Review Board

Stanford University

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

Medical IRB Study # 12659

Committee Name: Administrative Panel on Human Subjects in Medical Research

Seattle Children’s Hospital & Medical Center

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

Medical IRB Study # 12017

Committee Name: Institutional Review Board

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Hospital for Sick Children

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

REB File # 1000009409

Committee Name: Research Ethics Board (REB)

St. Michael's Hospital

Rare Genetic Disorders of the Airways: Cross-sectional Comparison of Clinical Features, and Development of Novel Screening and Genetic Tests

REB File # 10-367

Committee Name: Research Ethics Office

National Institute Allergy & Infectious Disease

Genetic Disorders of Mucociliary Clearance

Medical IRB Study # 06-I-0217

Committee Name: Institutional Review Board

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e-Table 1. Laterality Defects in Situs Ambiguus Participants with Classic PCD and without Classic PCD

ID	Age (yrs)	Situs	Classic PCD	Imaging	Cardiac apex	Gastric bubble	Spleen	Liver	Pulmonary	Other
PCD1	0.13*	SA + Complex CVM	Y	CXR, CT chest, echo, abdo US	R	L	L	R		DORV, MA, PS, L-TGA
PCD2	0.16*	SA + Simple CVM	Y	CXR, echo	R	R	unk	L		ASD
PCD3	1.8	SA + Complex CVM	Y	CXR, Chest CT, cardiac MRI, echo, abdo US	L	R	RPSP	L	LPI	DORV, LAI, ASD, VSD, I-IVC, TAPVR
PCD4	3.0	SA + Complex CVM	Y	CXR, CT sinus, echo, abdo US, BSS	R	R	RPSP	R	LPI	LAI, vascular ring, AVSD, HPV, RAA, TAPVR, B-SVC, malro
PCD5	4.7	SA + Simple CVM	Y	CXR, CT chest/sinus, echo, abdo US	R	R	R	L		D-TGA, ASD
PCD6	7.8	SA without cardiac defect	Y	CXR, echo, abdo US,	R	R	R	L	LPI	
PCD7	5.2	SA + Complex CVM	Y	CXR, Echo, abdo US	L	R	ASP	M		AVSD, CA, HPV, PS, RAA, TAPVR, malro
PCD8	6.4	SA without cardiac defect	Y	CXR, CT, echo, abdo US	R	R	RPSP	L	LPI	
PCD9	7.3	SA + Simple CVM	Y	CXR, CT chest, CT abdo, echo, Abdo US, BSS	L	R	R	L		VSD, malro

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PCD10	7.4	SA + Complex CVM	Y	CXR, CT chest, echo, abdo US, BSS	R	R	R	L		L-TGA, HPV, RAA, B-SVC
PCD11	8.1	SA without cardiac defect	Y	CXR, CT chest, abdo US, BSS	L	L	LPSP	M		I-IVC, malro
PCD12	8.1	SA + Simple CVM	Y	CXR, CT chest, echo	R	L	LPSP	M	LPI	ASD, RAA, TAPVR, malro
PCD13	8.5	SA + Simple CVM	Y	CXR, CT chest, echo, abdo US, BSS	R	R	R	L		B-SVC, ASD, VSD
PCD14	9.3	SA without cardiac defect	Y	CXT, CT chest, echo, BSS, abdo US	L	R	RPSP	L		malro
PCD15	10.7	SA + Simple CVM	Y	CXR, echo, abdo US	R	R	R	L		B-SVC, VSD
PCD16	11.1	SA without cardiac defect	Y	CXR, CT chest, echo	R	R	LPSP	M		RAA
PCD17	11.2	SA without cardiac defect	Y	CXR, CT chest, echo, BSS, abdo US	L	L	LPSP	R	LPI	I-IVC, malro
PCD18	11.4	Isolated possible laterality defect	Y	CXR, CT chest, echo	L	L	L	R		TOF
PCD19	11.5	SA + Complex CVM	Y	CXR, CT chest, echo, abdo US	L	M	LPSP	M	RPI	AVSD, DORV, PS, RAA, I-IVC, TAPVR, B-SVC
PCD20	12.4	SA + Complex CVM	Y	CXR, CT chest, echo	R	R	R	R		DORV, L-TGA, ASD, VSD, PS, B-SVC

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PCD21	12.9	SA + Complex CVM	Y	CXR, CT chest, echo	L	R	R	L		DORV, L-TGA, ASD, VSD, PS, RAA
PCD22	13.4	Isolated possible laterality defect	Y	CXR, CT chest	L	L	unk	R	LPI	
PCD23	14.0	Isolated possible laterality defect	Y	CXR, CT chest, echo	L	L	L	R		VSD
PCD24	15.6	SA without cardiac defect	Y	CXR, CT chest, CT abdo	R	R	RPSP	L		B-SVC
PCD25	16.0	Isolated possible laterality defect	Y	CXR, CT chest	L	L	L	R		I-IVC
PCD26	16.9	SA without cardiac defect	Y	CXR, CT chest, echo, BSS	R	R	RPSP	L	LPI	I-IVC, malro
PCD27	18.6	Isolated possible laterality defect	Y	CXR, CT chest, echo	L	L	L	R		VSD
PCD28	20.5	SA + Simple CVM	Y	CXR, CT chest, echo, BSS	R	R	R	L		ASD, VSD
PCD29	22.7	Isolated possible laterality defect	Y	CXR, CT chest/sinus, echo, abdo US, BSS	L	L	L	R		RAA

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PCD30	24.6	Isolated possible laterality defect	Y	CXR, CT chest, Echo, abdo US	L	L	LPSP	R		
PCD31	30.7	SA without cardiac defect	Y	CXR, CT chest, echo	L	R	RPSP	L		
PCD32	31.7	SA without cardiac defect	Y	CXR, CT chest/sinus, CT abdo, echo	R	R	RPSP	L		
PCD33	34.7	SA without cardiac defect	Y	CXR, CT chest, echo	R	L	LPSP	M		RAA, I-IVC
PCD34	35.8	SA without cardiac defect	Y	CXR, CT chest/sinus	R	R	RPSP	L		
PCD35	45.8	SA without cardiac defect	Y	CXR, ct chest, ct abdo, abdo us, BSS	L	0	RPSP	M		duplicated IVC
PCD36	55.2	Isolated possible laterality defect	Y	CXR, CT chest, echo	L	L	L	R		B-SVC
PCD37	58.0	SA without cardiac defect	Y	CXR, CT chest, CT abdo	L	R	ASP	R		I-IVC, bilateral livers, PKD
SA1	0.10*	SA + Complex CVM		CXR, CT chest, abdo US, echo	R	R	ASP	L		AVSD, HPV, PS, L-TGA, TAPVR, B-SVC

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SA2	0.50 *	SA without cardiac defect		abdo US, echo, BSS	L	R	RPSP	M		I-IVC, malro
SA3	0.76 *	SA + Complex CVM		CXR, CT chest, echo, abdo US, BSS	L	R	R	M	RPI	ASD, HPV, PS, I-IVC
SA4	0.77 *	SA + Complex CVM		CXR, echo, abdo US, BSS	L	R	ASP	M		AVSD, HPV, PS, L-TGA, TAPVR, malro
SA5	0.77 *	SA + Simple CVM		CXR, echo, abdo US, BSS	R	L	L	R		dextroposition, CA, PAPVR, I-IVC, L-SVC, RAA, malro
SA6	0.84 *	SA + Complex CVM		CXR, CT chest, echo	L	R	ASP	R		AVSD, ASD, VSD, Atrial SI, DORV, D-TGA, TAPVR, B-SVC
SA7	0.88 *	SA + Simple CVM		CXR, echo	R	R	unk	unk		L-TGA, I-IVC, RAA
SA8	1.1	SA + Simple CVM		CXR, CT chest, echo, abdo US, BSS	R	L	L	R		dextroposition, dextrorotation, ASD, B-SVC
SA9	1.6	Isolated possible laterality defect		CXR, echo, abdo US	R	L	L	R		Isolated dextrocardia
SA10	1.6	SA + Complex CVM		CXR, echo, BSS	R	R	RPSP	M		L-TGA, ASD, HPV, I-IVC, malro
SA11	2.3	SA without cardiac defect		CXR, echo, abdo US, BSS	R	R	RPSP	L		I-IVC, B-SVC, malro
SA12	3.0	SA without cardiac defect		CXR, echo, abdo US	R	R	RPSP	M		

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SA13	3.2	Isolated possible laterality defect		CXR, CT chest, BSS	L	L	unk	R		ASD, VSD
SA14	3.3	SA + Simple CVM		CXR, echo, BSS	L	L	unk	R		B-SVC, D-TGA, ASD
SA15	3.3	Isolated possible laterality defect		CXR, CT chest/sinus, echo, BSS	L	L	L	R		PAPVR
SA16	3.6	SA + Simple CVM		CXR, CT chest, echo, BSS	L	L	L	R		D-TGA, PS, hydrocephalus
SA17	4.0	SA + Complex CVM		CXR, echo, abdo US, BSS	R	L	LPSP	R		ASD, VSD, PS, L-TGA, I-IVC, malro
SA18	4.9	SA + Simple CVM		CXR, CT chest, echo, BSS	R	R	RPSP	L		ASD, VSD, I-IVC, B-SVC
SA19	5.3	SA without cardiac defect		CT chest, CT abdo, echo, BSS	L	R	RPSP	M		I-IVC
SA20	5.6	SA + Simple CVM		CT chest, Sinus xray, echo	R	R	R	L		D-TGA, coarct
SA21	5.9	SA + Simple CVM		CXR, CT chest, echo, BSS, abdo US	L	R	RPSP	M		OVR AOR, VSD, I-IVC
SA22	6.2	SA + Simple CVM		CXR, Echo	R	R	unk	L		OVR AOR, VSD, D-TGA, B-SVC
SA23	6.5	SA + Simple CVM		CXR, echo, abdo US	R	R	R	L		ASD

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SA24	6.8	SA without cardiac defect		CXR, CT chest, echo, BSS	R	R	LPSP	L		malro
SA25	7.3	SA + Complex CVM		CXR, CT chest/sinus, echo, abdo US	R	L	ASP	R		DORV, AVSD, ASD, L-TGA, PS
SA26	7.4	Isolated possible laterality defect		CXR, CT chest, echo, BSS	L	L	unk	R		D-TGA
SA27	8.0	SA + Complex CVM		CXR, CT chest/sinus, echo, abdo US, BSS	L	L	ASP	M	RPI	AVSD, HPV, PS, D-TGA, RAA, I-IVC, TAPVR, B-SVC, malro
SA28	8.1	SA + Complex CVM		CXR, CT chest, echo, abdo US	L	R	ASP	M		AVSD, DORV, PS, L-TGA, RAA, TAPVR, B-SVC, malro
SA29	8.4	Isolated possible laterality defect		CXR, CT chest, echo	L	L	L	R		TOF
SA30	8.7	Isolated possible laterality defect		CXR, CT chest, echo	L	L	L	R		PS
SA31	9.2	SA + Complex CVM		CXR, CT chest, echo, abdo US	R	R	R	L		DORV, PA, MA, B-SVC
SA32	10.3	SA + Simple CVM		CXR, CT chest, echo	R	R	R	L		VSD
SA33	10.8	SA + Simple CVM		CXR, CT chest, echo	L	L	LPSP	R	LPI	AVSD, I-IVC, B-SVC, malro, biliary atresia

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SA34	12.0	SA + Complex CVM		CXR, echo, abdo US, cardiac MRI, cath	L	R	ASP	L		AVSD, HPV, PS, D-TGA, I-IVC, TAPVR, B-SVC
SA35	15.2	SA + Simple CVM		CXR, CT chest/sinus, echo, abdo US	L	L	LPSP	R	LPI	VSD, PS, I-IVC, malro, absent distal pancreas
SA36	15.9	SA + Simple CVM		CXR, CT chest/sinus, echo, BSS	M	L	L	R		B-SVC
SA37	16.3	Isolated possible laterality defect		CXR, CT chest, echo	L	L	L	R		TAPVR
SA38	25.1	SA + Complex CVM		CXR, CT chest/sinus, echo, abdo US, BSS	L	L	ASP	M	RPI	AVSD, CA, HPV, PS, D-TGA, TAPVR, malro, annular pancreas, duodenal atresia
SA39	26.3	SA without cardiac defect		CT chest, CXR, echo	L	R	RPSP	M		
SA40	34.7	SA without cardiac defect		CXR, CT chest, CT abdo	L	R	RPSP	L	LPI	I-IVC
SA41	38.8	SA without cardiac defect		CXR, CT chest, CT abdo, echo, abdo US, spleen scan	R	R	RPSP	L	LPI	RAA, B-SVC, Coarc

*Participants under 1 year of age not included in data analysis.

Abdo US – abdominal ultrasound, ann panc – annular pancreas, AVSD - atrioventricular septal defect, ASD - atrial septal defect, ASP – asplenia, bil atr – biliary atresia, BSS – barium swallow study, B-SVC - bilateral superior vena cava, coarc - coarctation of aorta, CA - common atrium, cath – cardiac catheterization, CHHD- classic heterotaxic heart defect,, CXR – chest x-ray, D-TGA - dextro transposition of great arteries, DORV - double outlet right ventricle, Echo – echocardiogram, HPV - hypoplastic ventricle, I-IVC -

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interrupted inferior vena cava, L – left, LAI - left atrial isomerism, L-TGA - levo transposition of great arteries, LPSP – left polysplenia, LPI – left pulmonary isomerism, M – mesocardia, malro - intestinal malrotation,, Mid – midline, OVR AOR - overriding aorta, PAPVR – partial anomalous pulmonary venous return, PS - pulmonary stenosis/atresia, PSP – polysplenia, R – right, RAA - right aortic arch, RPSP – right polysplenia, RPI – right pulmonary isomerism, TAPVR - total anomalous pulmonary venous return, TOF - tetralogy of Fallot, unk – unknown status, VSD – ventricular septal defect.

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