

Supplementary Table S1 Clinical features of the six patients with spontaneous pubertal development (SMI-6) and of seven patients with primary amenorrhea (PAI-7) representative of the full PA group.

Case	Age (y)	Karyotype	Statural growth		Skeletal anomalies							Soft tissue anomalies			Visceral anomalies		Others	
			AH (cm)	TH (cm)(cm)	GHt	SM	CV	WA	HP	SN	WN	LH	LE	ND	Cardiac ^a	Renal ^b		PN
SM1	32	45,X	145.1	169.5	+	-	+	-	+	-	-	-	-	-	Rope accessory tendon in the left ventricle; mitral valve prolapse	-	+	
SM2	18	45,X	140.5	154	-	+	+	-	+	+	-	+	-	-	Mild ectasia of the aortic bulb; tricuspid regurgitation	Right renal pelvis hypotonia and left cortical cysts	+	
SM3	15	45,X/46,XX	144 ^c	158.75	+	-	+	-	+	-	-	-	-	+	Mild ectasia of the aortic bulb	-	+	
SM4	19	45,X/46,XX	158.1	161.15	+	-	-	-	+	-	-	-	-	-	-	-	-	+
SM5	15	45,X/46,XX	157 ^c	159.5	-	-	+	-	-	-	-	-	-	-	-	-	-	+
SM6	15	45,X/46,XX	150 ^c	159.6	+	-	-	-	+	-	-	-	-	-	Mild ectasia of the aortic bulb	-	+	
PA1	46	45,X	130.4		-	-	+	-	+	+	+	+	+	+	Rope accessory tendon in the left ventricle; mitral valve prolapse; anomalous venous return; mild tricuspid regurgitation	-	+	
PA2	43	45,X	149.6	163.5	-	+	+	-	+	-	-	-	-	-	Mitral valve prolapse	-	+	
PA3	24	45,X	159.8	157.45	+	-	+	-	+	-	-	-	-	-	-	-	-	+
PA4	35	45,X/46,XX	142	148.5	+	+	+	-	-	-	-	-	-	-	Rope accessory tendon in the left ventricle	Horseshoe kidney	+	
PA5	44	45,X/ 46,X,i(Xq)	136.7	165	+	-	+	-	+	+	-	-	-	-	Mitral valve prolapse	-	+	
PA6	15	45,X/ 46,X,i(Xq)	152.9 ^c	165.1	+	-	+	-	+	-	-	-	+	+	Ectasia of the aortic bulb with aortic regurgitation	-	+	
PA7	33	45,X/ 46,X,i(Xq)	157.4	167.15	+	+	+	-	+	-	-	-	-	-	Atrial septal defect; mitral valve prolapse; rope accessory tendon in the left ventricle	-	+	

AH, actual height; CV, cubitus valgus; GHt, growth hormone therapy; HP, high-arched palate; LE, lymphedema; LH, low posterior hairline; ND, nail dysplasia; PN, multiple pigmented nevi; SM, short metacarpal; SN, short neck; TH, target height; WA, wrist abnormality; WN, webbed neck.

^aCardiac anomalies were evaluated by echocardiography.

^bRenal anomalies were evaluated by renal ultrasound.

^cAt the last follow-up the patients had not yet reached their final height.