

Supplementary data.

Patients with LVOTO associated with NCA

a) Patients with a *de novo* chromosomal aberration (n=11)

Db no.	non-cardiac anomalies	Cardiac defect	chromosomal aberration	location* (Mb)
264	Rieger's anomaly + hearing loss+ FD	BAV/AVS	del 6p25.3	0.11-2.25
85	ankyloglossus	COA + VSD	del 15q11.2	18,7-20,9
203	thyroid agenesis + cryptorchism +MR	BAV/AVS + MVS	del 3q29	197,0-198,9
131	microcephaly + hearing loss + FD	BAV + RAA	del 22q11**	
257	microcephaly + FD	BAV + VSD	mos 47,XX r(7)(p22q32)	
325	microcephaly + MR + micropenis	BAV	del 2q24.3q32.1	168,36-184,63
341	Turner syndrome	COA + BAV	del Xp11.23	
344	Turner syndrome	COA + BAV	45,X	
169	Turner syndrome	COA + BAV	45,X	
343	Turner syndrome	COA	45,X	
342	Turner syndrome	BAV	45,X	

b) Patients with normal karyotype/WGA (n=29)

Db no.	non-cardiac anomalies	Cardiac defect	specific test
13	microcephaly + anotia + lens luxation	COA + VSD	
141	microcephaly + MR	COA + VSD	
288	microcephaly + FD + MR	COA + VSD	
196	microcephaly + hydrocephaly	COA	
235	anotia, absent thumb	HLHS	
95	Coffin Siris syndrome	COA	
321	CHARGE syndrome	HLHS	mutation CHD7
330	CHARGE syndrome	COA + VSD	mutation CHD7
126	Cleft lip and palate	COA + BAV + MVS	
137	esophagus atresia + cataract	COA+VSD	

139	omphalocele	COA
195	hypospadias + genital ambiguity + FD	HLHS
336	pectus excavatum + FD	MVS + VSD
315	Sprengel's deformity	BAV/AVS
9	preauricular tags	BAV
40	hypoplasia + malposition Dig. I	BAV +VSD
43	cyst in mandible	HLHS + COA
53	scoliosis, vertebral anomalies	COA + BAV + VSD
130	unilateral dysplastic ear	BAV/AVS
145	FD	HLHS
146	contractures, skin tags, hypertelorism	AVS + VSD
201	horseshoe kidney + FD	HLHS
247	accessory nipple + FD	COA + BAV
292	polydactyly	BAV/AVS
228	FD	BAV/AVS
88	MR + radio-ulnar synostosis	BAV/AVS
277	MR + syndactyly	COA + VSD + PDA
205	hydrops foetalis	BAV/AVS
247	accessory nipple + FD	BAV/AVS

AVS aortic valve stenosis; BAV bicuspid aortic valve; CLP cleft lip and palate; COA coarctation of the aorta;

Db no. database number; del deletion; dig I digitus I; FD facial dysmorphism; HLHS hypoplastic left heart syndrome; LVOTO left ventricular outflow tract obstructions (definition see text); MR mental retardation; MVS mitral valve stenosis; NCA non-cardiac anomalies; PDA persistent ductus arteriosus; RAA right sided aortic arch; VSD ventricular septal defect; WGA whole genome array; * location is defined as distance from p-telomere in Megabases; **diagnosed by FISH with cosmids on D22S183 and D22S111 and a control probe on 22q13.