Table S1

Duplication break points	Cardiovascular defect	Classification	Sex	Inherited	Reference
A-D	TOF	OFT	Male	Unknown	CHOP
A-D	TOF	OFT	Female	Inherited (mother)	
A-D	PTA	OFT	Female	De novo	N=24
A-D	HLHS	HLHS	Male	Inherited (father)	
A-D	HLHS	HLHS	Female	Inherited (mother)	
A-D	ASD	ASD	Female	Inherited (father)	
A-D	ASD	ASD	Male	De novo	
A-D	ASD	ASD	Male	Inherited (mother)	
A-D	VSD	OFT	Male	Inherited (father)	
A-D	VSD	OFT	Male	Unknown	
A-D	VSD	OFT	Male	Inherited (mother)	
A-D	ASD and VSD	OFT/ASD	Male	Unknown	
A-D	ASD and VSD	OFT/ASD	Female	Unknown	
A-D	TAPVC with ASD	ASD	Male	De novo	
A-D	TAPVC with ASD	ASD	Male	Unknown	
A-D	PV stenosis	OFT	Female	Unknown	
A-D	PV stenosis	OFT	Male	De novo	
A-B	ASD	ASD	Male	Inherited (mother)	
A-B	ASD	ASD	Female	Unknown	
FISH	PFO with PA stenosis	OFT	Female	Inherited (father)	
FISH	PV stenosis	OFT	Female	Unknown	
FISH	PV stenosis	OFT	Male	Inherited (father)	
FISH	TOF	OFT	Female	Unknown	
FISH	VSD	OFT	Male	Unknown	
A-D	PTA	OFT	Female	De novo	
A-D	VSD	OFT	Female	Inherited (father)	26297018
A-D	ASD	ASD	Female	De novo	
A-D	PTA	OFT	Female	Inherited (father)	16490798
A-B	Right aortic arch	OFT	N/A	Inherited (father)	25118001
A-B	TGA + VSD	OFT	N/A	Inherited (father)	N=26 total cases
A-C	HLHS	HLHS	N/A	Unknown	
A-D	HLHS	HLHS	N/A	Inherited (mother)	
A-D	PV stenosis + VSD	OFT	N/A	Unknown	
A-D	OFT does not specify	OFT	N/A	Unknown	
A-D	OFT does not specify	OFT	N/A	Unknown	24098474
A-D	OFT does not specify	OFT	N/A	Unknown	
A-D	PTA, VSD, tricuspid atresia, IAA	OFT	Female	Inherited (mother)	21473936
A-D	DORV +HLHS+ ASD	HLHS, OFT, ASD	Female	Inherited (father)	16761289 n=3
A-D	ASD	ASD	Female	Inherited (father)	23159380
A-D	VSD	OFT	Female	Inherited (mother)	18076674 n=8
A-B	PTA	OFT	Male	Inherited (mother)	23059467
A-C	HLHS + PV stenosis	HLHS, OFT	Female	De novo	24451223
A-C	Aorta hypoplasia + VSD	OFT	N/A	Inherited	26715944
1.4 MB include Tbx1	VSD	OFT	N/A	Inherited (father)	26625662
A-D	ASD	ASD	Female	De novo	25376777
A-D	Complex CHD		Male	Unknown	21849782
A-D	PTA	OFT	N/A	Inherited (father)	17384091
FISH p250 kit	TGA	OFT	Male	De novo	25387403
FISH TUPLE1	TGA + VSD	OFT	Male	Inherited (father)	21199755
FISH TUPLE1	TOF	OFT	Male	Unknown	14526392
FISH TUPLE1	HLHS + IAA	HLHS	Male	Unknown	N=13
FISH TUPLE1	TGA	OFT	Female	Inherited (mother)	20020400
FISH TUPLE1	chronic aortic regurgitation, mild mitral and tricuspid regurtitation	Valve defect	Male	Inherited (father)	15099348
FISH	Mitral valve defect	Valve defect	Female	Inherited (father)	17455106
FISH TUPLE1	TOF	OFT	Female	Inherited (father)	15845171
FISH TUPLE1	HLHS	HLHS	Male	De novo	
FISH TUPLE1	TOF	OFT	Female	Unknown	15800846 n=7
FISH TUPLE1	HLHS	HLHS	Male	Unknown	

Table S1. **Detailed information of all individuals found with *TBX1* included in duplication break points.** Type of duplication, cardiac defect and category of the CHD, sex and inheritance (when these information was available). Human LCR22-A-D duplication is indicated as A-D; LCR22-A-B duplication is indicated as A-B; LCR22A to LCR22C is indicated as A-C. *TBX1* lies between duplicated region A-B and is among the duplicated region in each of these listed cases. Abbreviations: Tetralogy of Fallot (TOF); persistent truncus arteriosus (PTA); hypoplastic left heart syndrome (HLHS); atrial septal defect (ASD); ventricular septum defect (VSD); pulmonary valve (PV); pulmonary artery (PA); transposition of great arteries (TGA); double outlet right ventricle (DORV); interrupted aortic arch (IAA); total anomalous pulmonary venous connection (TAPVC); patent foramen ovale (PFO); outflow tract (OFT).