

Congenital heart disease	Extracardiac anomaly	Genetic diagnosis
TGA	CCAM	Amniocentesis karyotype: normal
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TGA	Abdominal situs inversus	Genetic test not performed
TGA	Bilateral cleft lip and palate	Postnatal karyotype: normal
Truncus arteriosus	Left multicystic kidney	Amniocentesis karyotype: normal
Tetralogy of Fallot	Ectopia cordis	Amniocentesis karyotype: normal
Tetralogy of Fallot	Bilateral talipes equinovarus	Lost to follow-up
Tetralogy of Fallot	Epilepsy, global developmental delay, central hypotonia, central apnoea, nystagmus, hepatomegaly, supernumerary nipples	Postnatal chromosomal microarray: benign familial copy variants WES: homozygous mutation in PIGN causing congenital disorder of glycosylation
Tetralogy of Fallot	1) Spinal angulation at thoracic level 2) Dismorphic facies with micrognathia	Amniocentesis karyotype: normal
Tricuspid atresia	Right ulnar dysplasia/hypoplasia with intrauterine growth restriction	Amniocentesis karyotype: 46,XX,der(21)ins(21;?)(q11.2;?)
UVH	Abdominal situs inversus	Lost to follow-up
AVSD	Inferior cerebellar vermian hypoplasia	Postnatal karyotype: trisomy 21
AVSD	Bilateral adducted fourth toe	Postnatal CMA: duplication of 444 kb in 15q13.3 encompassing part of <i>CHRNA7</i> gene. Known to have variable penetrance and not associated with AVSD. No parental CMA.
Large ventricular septal defect	Right talipes equinovarus	Postnatal karyotype: normal. FISH: 22q deletion detected

AVSD: atrioventricular septal defect, CCAM: congenital cystic adenomatoid malformation, CMA: chromosomal microarray analysis, FISH: fluorescence in-situ hybridisation, TGA: transposition of arteries, UVH: univentricular hearts, WES: whole exome sequencing