Supplementary Material Table S1. Shows the details of pathogenic copy number variations in 38 CHD fetuses.

| case | Cardiac and Extra Cardiac Phenotype | Chr | pCNVs | Type | Size (Mb) | Syndromes | Major genes included | Pattern of Inheritance | Outcomes |
|---|---|---|---|---|---|---|---|---|---|
| 1 | VSD，endocardial fibroelastosis, MCDK | 1 | 849,466-5,747,184 | del | 4.8 | 1p36 deletion syndrome | *PRDM16, SKI* | / | TP, Autopsy [a] |
| 2 | TOF，FGR，limb abnormality | 5 | 36,069,681-40,557,872 | del | 4.4 | Cornelia de Lange | ***NIPBL*** | / | TP |
| 3 | VSD，spina bifida | 6 | 163,732,938-170,914,297 | del | 7.1 | 6q terminal deletion syndrome | ***DLL1*** | / | LTF |
| 4 | CoA，FGR | 7 | 72,653,992-74,154,209 | del | 1.5 | WBS | *ELN, LIMK1* | / | LTF |
| 5 | CAT, VSD, horseshoe kidney | 8 | 160,101-7,120,108 | del | 6.9 | | *CSMD1, ANGPT2* | / | TP, Autopsy [a] |
| 6 | HRHS，VSD，callosal dysplasia，SUA | 10 | 98,561,770-105,586,041 | dup | 7.0 | | *LBX1, BTRC, POLL* | / | TP, Autopsy[b1] |
| 7 | HLHS，hemivertebra | 11 | 12,674,297-13,486,847 | del | 8.8 | | ***FLI1*** | / | TP, Autopsy [a] |
| 8 | Single ventricle, DORV, PS | 11 | 81,354,834-106,214,708 | del | 24.8 | | *FZD4* | / | TP |
| 9 | Aortic stenosis, VSD, hydrocephalus | 12 | 103,403,037-133,777,562 | dup | 30.3 | Noonan syndrome 1 | ***PTPN11, TBX5*** | / | TP |
| 10 | VSD，CCAM | 16 | 15,438,164-18,172,468 | dup | 2.7 | 16p13.11 recurrent microduplication | *MYH11* | / | LTF |
| 11 | TOF，hyperechogenic bowel, ventriculomegaly | 17 | 16,736,262-20,417,235 | del | 3.6 | SMS | *RAI1* | / | TP, Autopsy [a] |
| 12 | VSD，osteogenic dysplasia | 17 | 14,077,970-15,482,833 | del | 1.4 | HNPP | *PMP22* | / | LTF |
| 13 | PA，VSD，RAA，choroid plexus cyst | 21 | 33,623,086-35,352,874 | del | 1.7 | Tokita-Kim syndrome | *RUNX1, SON, SYNJ1* | / | TP |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| 14 | VSD, PS, tricuspid valve dysplasia | 17 | 87,009–1,184,534 | del | 1.1 | MDLS | *LIS1* | / | TP， | Autopsy [c] |
| | | 22 | 18,919,942–21,440,514 | del | 2.5 | 22q11.2 DS | *TBX1* | | | |
| 15 | d-TGA, VSD | 22 | 18,901,424-21,790,211 | del | 2.8 | 22q11.2 DS | | / | TP | |
| 16 | VSD, cor triatriatum, mitral valve dysplasia, SUA | 22 | 18,984,187-21,927,646 | dup | 2.9 | 22q11.2 microduplication syndrome | | / | TP， | Autopsy [a] |
| 17 | TOF(APVS), VSD | 22 | 18,916,842-21,798,907 | del | 2.8 | 22q11.2 DS | | denovo | TP | |
| 18 | VSD, aortic stenosis, hydramnios | 22 | 18,636,749-21,800,471 | del | 3.1 | 22q11.2 DS | | denovo | TP | |
| 19 | TOF | 22 | 18,644,790-21,465,659 | del | 2.8 | 22q11.2 DS | | denovo | TP， | Autopsy [c] |
| 20 | RAA， APLPA | 22 | 16,888,899-20,725,309 | del | 3.8 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 21 | VSD, PS | 22 | 18,636,749-21,800,471 | del | 3.1 | 22q11.2 DS | | denovo | TP | |
| 22 | VSD, CoA | 22 | 18,648,855-21,800,471 | del | 3.1 | 22q11.2 DS | | / | TP | |
| 23 | DORV, VSD, PA | 22 | 18,939,748-21,721,712 | del | 2.7 | 22q11.2 DS | | denovo | TP， | Autopsy [c] |
| 24 | RAA， VSD, left superior vena cava | 22 | 19,004,071-21,800,471 | del | 2.7 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 25 | RAA， VSD | 22 | 18,901,201-21,480,190 | del | 2.5 | 22q11.2 DS | | / | TP | |
| 26 | TOF | 22 | 18,906,341-21,460,123 | del | 2.5 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 27 | TOF | 22 | 18,978,201-21,926,534 | del | 2.9 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 28 | TOF, RAA， absence of ductus arteriosus | 22 | 18,636,750-21,800,471 | del | 3.1 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 29 | VSD, CoA | 22 | 18,648,856-21,800,471 | del | 3.1 | 22q11.2 DS | | / | TP， | Autopsy[b2] |
| 30 | TOF, RAA, thymus aplasia | 22 | 18,648,856-21,058,888 | del | 2.4 | 22q11.2 DS | | / | TP， | Autopsy [a] |
| 31 | IAA, type A, aortic stenosis, ARSA | 22 | 18,648,856-21,800,471 | del | 3.1 | 22q11.2 DS | | / | TP， | Autopsy [c] |
| 32 | TOF | 22 | 18,882,825-21,796,237 | del | 3.1 | 22q11.2 DS | | / | TP | |
| 33 | TOF (PA-VSD) | 22 | 18,927,431-21,427,431 | del | 2.5 | 22q11.2 DS | | / | TP | |
| 34 | Single ventricle, single atrium, CAT | 22 | 46933489-51219152 | del | 4.2 | PHMDS | *SHANK3* | denovo | TP | |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| 35 | TAPVC, accessory ear, micrognathia | 22 | 16050230-18679614 | dup | 2.6 | CES | | / | TP，Autopsy [a] |
| 36 | RAA | X | 2,703,661-6,609,379 | del | 3.9 | | *ARSL* | denovo | Birth |
| 37 | TOF (PA-VSD), RAA, left superior vena cava, SUA | X | 168,552-55,508,714 | Del | 55.3 | LWD, Xq28 duplication | *SHOX, DMD, USP9X, GATA1, PIGA MECP2* | / | TP，Autopsy [a] |
| | | | 55,509,386-155,233,098 | dup | 99.7 | | | | |
| 38 | VSD, holoprosencephaly, facial abnormality | X | 134,743,723-155,043,723 | dup | 20.3 | Xq28 duplication | *MECP2* | / | TP |

MCDK，multicystic dysplastic kidney; SUA，single umbilical artery; CoA, coarctation of aorta; CAT, common arterial trunk; CCAM，congenital cystic adenomatoid malformation; APVS, absent pulmonary valve syndrome; APLPA, anomalous origin of left pulmonary from the ascending aorta; ARSA, aberrant right subclavian artery; DS: deletion syndrome; del: deletion; dup: duplication; WBS: Williams-Beuren Syndrome; SMS: Smith-Magenis syndrome; HNPP: Hereditary Liability to Pressure Palsies; MDLS: Miller-Dieker lissencephaly syndrome; PHMDS: Phelan-McDermid syndrome; CES: Cat eye syndrome; LWD: Leri-Weill dyschondrostsosis; LTF, lost to follow-up; TP, terminal pregnancy.

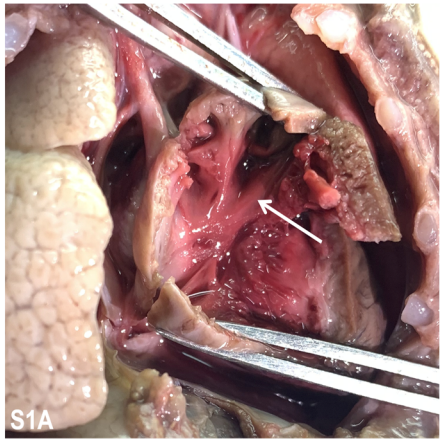
[a] autopsy results are consistent with prenatal ultrasound.

[b1] autopsy revealed repeated bladder and anal atresia, others were consistent with prenatal ultrasound.

[b2] autopsy result is IAA, type B.

[c] autopsy also found thymus hypoplasia/aplasia.

Bold font is the genes discussed in the article.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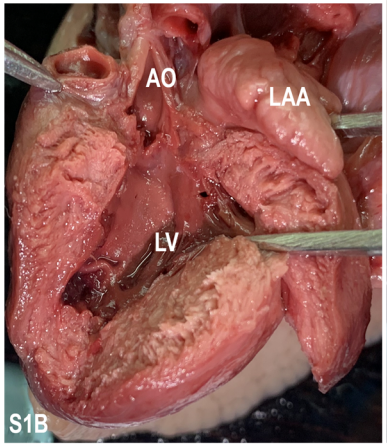
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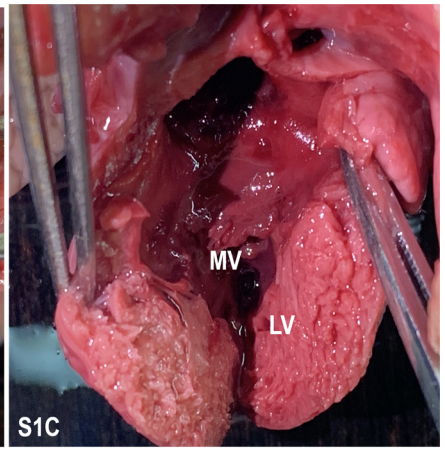
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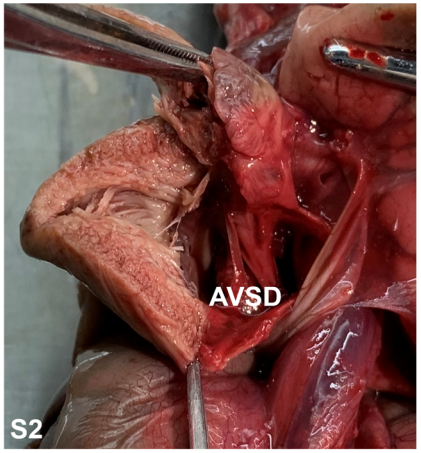
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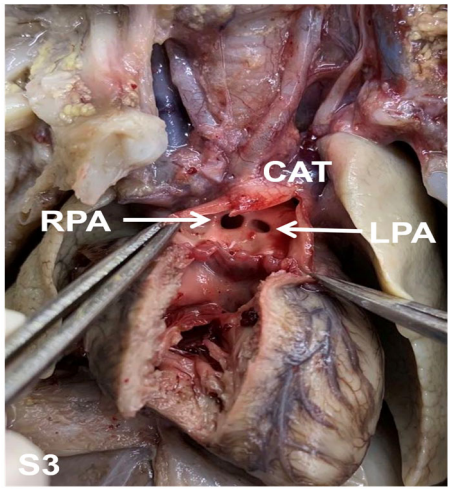
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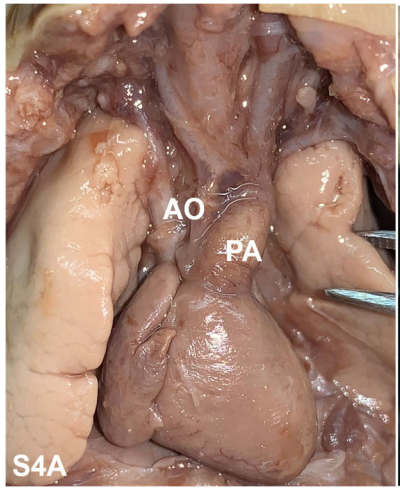
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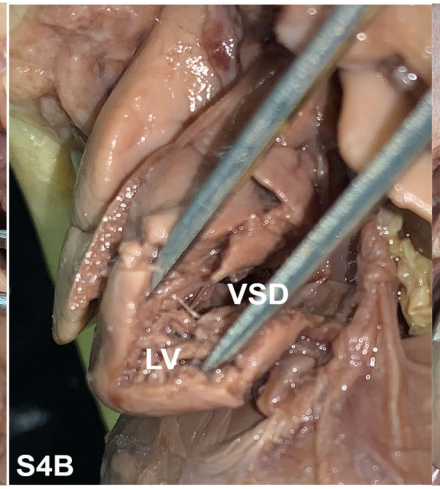
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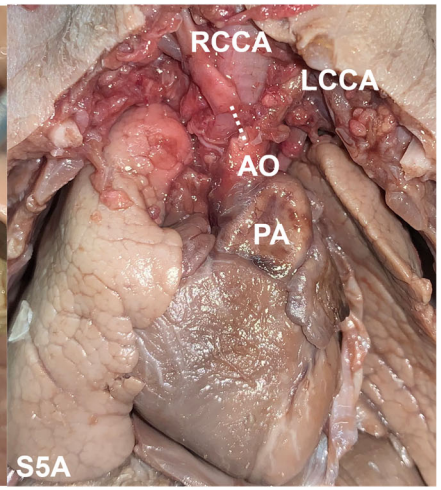
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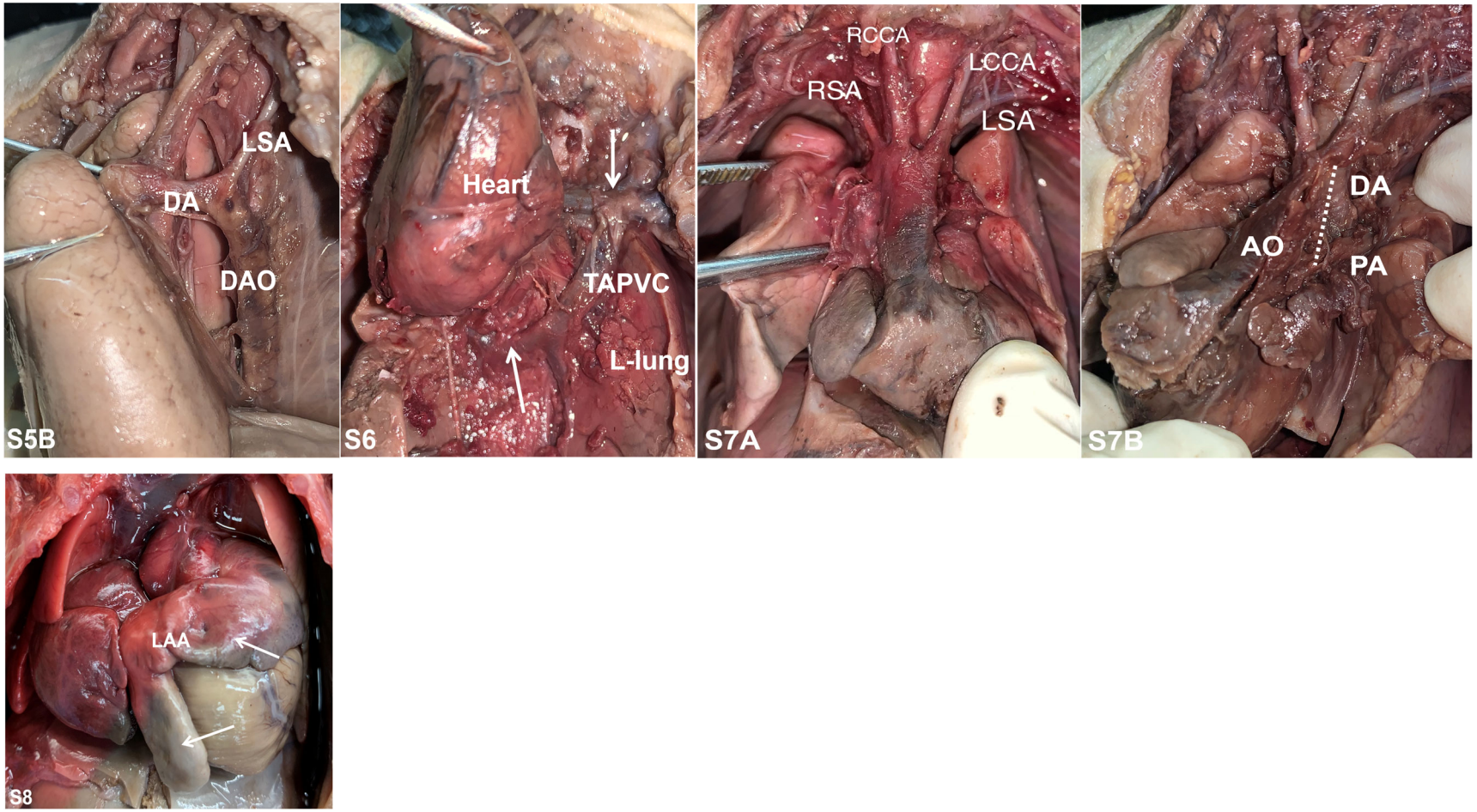


**Figure S1-S8 shows the autopsy of CHD fetus.**

Fig S1, mitral valve dysplasia; S1A, arrow shows no foramen ovale; S1B, LV and LAA thickening, stenosis aorta; S1C, mitral valve dysplasia, CNV result was VUS; Fig S2, complete type AVSD, trisomy 18; Fig S3 shows case 5 in table 2, autopsy results show CAT (type II); Fig S4 shows

case 7 in table 2; S4A, aorta dysplasia; S4B, LV is very small, VSD can be seen. Fig S5 shows case 29; S5A, only two blood vessels are sent out in ascending aorta; S5B, LSA is sent from descending Ao. Figure S6 shows case 35, and the arrows show common vena cava and vertical vein, respectively; Figure S7 shows case 37; S7A, RAA with mirror image branching; S7B, PA atresia, left and right pulmonary arteries supplied by DA. Dotted line, blood vessel accidentally broken; LAA, left atrial appendage; Ao, aorta; PA, pulmonary artery; LV, left ventricle; MV, mitral valve; CAT, common arterial trunk; LCCA, left common carotid artery; RCCA, right common carotid artery; LSA, subclavian artery; DA, ductus arteriosus. Fig S8, prenatal diagnosis of abnormal cardiac function, autopsy found a huge LAA, CNV result was VUS.