Prenatal Diagnosis and Successful Palliation of Absent Aortic Valve with Hypoplastic Left Heart Syndrome: A Case Report and Review of Literature

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Abstract

Introduction  Congenital absence of the aortic valve leaflets is a rare association with hypoplastic left heart syndrome (HLHS).

Case  A 37-year-old pregnant woman was referred for fetal evaluation of possible HLHS at 22 weeks of gestation. The fetal echocardiogram (ECHO) was remarkable for a hypoplastic left atrium, nearly atretic mitral valve, small left ventricle, and a hypoplastic aortic valve with severe aortic insufficiency. A female infant was born at term and postnatal ECHO confirmed the above findings. In addition, there was complete absence of the aortic valve leaflets. The patient underwent Norwood’s procedure at day 5 of life with atrial septectomy, over-sewing of the aortic valve annulus, and a 4 mm Sano’s shunt between the right ventricle and the main pulmonary artery. She tolerated this surgery well and subsequently underwent a bidirectional Glenn’s procedure at 8 months of life.

Conclusion  Prenatal diagnosis of absent aortic valve should be suspected in the presence of severe aortic insufficiency in the fetal ECHO. Early postnatal intervention is critical as those patients are likely to deteriorate quickly. The over-sewing of the aortic valve may be important to prevent coronary steal and myocardial hypoperfusion which could potentially be detrimental.

Absence of the aortic valve (AAV) is a very rare congenital heart defect, unlike the absence of pulmonary valves. AAV is usually associated with other cardiac and non-cardiac anomalies. Most of the previously reported cases have been in spontaneously aborted fetuses, underscoring the high mortality of this heart defect.

Case

A 37-year-old pregnant female (Gravida 5, Para 3) was referred for fetal echocardiogram (ECHO) due to concerns of hypoplastic left heart (HLH) on anatomy ultrasound scan. Fetal ECHO at 22 weeks was significant for a hypoplastic left atrium, nearly atretic mitral valve, small nonapex forming left ventricle (LV), hypoplastic aortic valve with severe aortic insufficiency (► Fig. 1). Serial fetal ECHOs showed the same findings throughout pregnancy. No evidence of hydrops was seen. At 39-week gestation, a female infant was delivered with a birth weight 3.2 kg and Apgar’s scores of 8 and 8 at 1 and 5 minutes, respectively. Prostaglandin E-1 (0.05 mcg/kg/min) infusion was started within an hour of delivery. Postnatal ECHO findings (► Fig. 2, ► Video 1) showed HLH variant with absent aortic valve and severe aortic insufficiency, a large PDA, moderate sized atrial septal defect (ASD), and poor left ventricular posterior wall function. The LV was diffusely echogenic but there was no clear endocardial fibro elastosis.

Keywords

► fetal echocardiography (ECHO)  
► absent aortic valve  
► hypoplastic left heart syndrome

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There was decimal mitral inflow and a mild tricuspid valve insufficiency. The right ventricle was globular with a normal function. The coronary anatomy was normal but the flow was difficult to assess. Since the patient had no dysmorphic features or any extra cardiac anomaly, no genetic testing was done. The patient successfully underwent the first stage Norwood’s procedure on the 5th day of life. The procedure included atrial septectomy, an end to side main pulmonary artery to aorta (Damus-Kaye-Stansel) anastamosis, PDA ligation, over-sewing of the aortic valve, and the placement of a 4 mm Sano’s shunt between the right ventricle and the main pulmonary artery. Subsequently, she underwent the bidirectional Glenn’s procedure at 8 months of life and is currently doing well and maintaining her O₂ saturations in the mid-80s on room air.

Video 1

A parasternal long axis view showing a hypoplastic left ventricle and an absent aortic valve with severe regurgitation. Online content including video sequences viewable at: www.thieme-connect.com/products/ejournals/html/10.1055/s-0038-1677480.

Fig. 1  Fetal ECHO "4-chamber view" showing a hypoplastic left ventricle and left atrium (left) and color Doppler’s image showing aortic regurgitation (right).

Fig. 2  Postnatal ECHO showing a hypoplastic left ventricle seen in 4-chamber view (left) and aortic regurgitation seen in parasternal long axis view (right).
Discussion

Absent aortic valve (AAV) is a rare congenital heart defect with our case being the 26th that was reported in literature. It is usually associated with other congenital anomalies including HLH, double outlet right ventricle (DORV), mitral atresia and absent, or dysplastic pulmonary valve. The clinical presentation may vary but mainly includes cyanosis, respiratory distress and cardiomegaly in the majority of reported cases. Table 1 shows a review of previously

| Case no., gender | Diagnosis age and method | Other CHD | Noncardiac anomalies | Clinical presentation | Outcome | Ref. |
|------------------|--------------------------|----------|----------------------|-----------------------|---------|------|
| 1, M             | 36 h, autopsy            | DORV, enlarged RA and RV, ASD, PDA, dysplastic LV, hypoplastic MV and LV | Accessory spleen      | Severe cyanosis and cardiomegaly at 36 h | Death at 2 d | 7    |
| 2, M             | 32 wk, fetal ECHO + autopsy | DORV, common AV canal, hypoplastic MV and LV | Absent R SVC, anomalous L SVC, splenic nodules, gut malrotation | Hydrops, severe polyhydramnios, RD | Death at 20 h | 8    |
| 3, M             | 24 h, ECHO + autopsy     | Hypoplastic MV and LV, EFE of LV | None | Cyanosis, RD, tachycardia | Death at 8 d | 9    |
| 4, M             | 4 h, ECHO + autopsy      | Hypoplastic LA, EFE of LV, anomalous RSA, dysplastic TV and PV, PDA | 3 lobes in left lung and horseshoe kidneys | Severe cyanosis and RD | Death at 4 d | 10   |
| 5, F             | 1 d, autopsy              | ASD, VSD, Interrupted aortic arch, anomalous RSA | DiGeorge’s syndrome (absent thymus, PTH glands) | RD, cardiomegaly | Death at 36 h | 11   |
| 6, M             | 12 h, ECHO + autopsy     | MV atresia, Dysplastic PV, PDA, normal LV | ND | RD, cyanosis | Death at 24 h | 12   |
| 7, M             | 11 h, ECHO + autopsy     | Dysplastic MV, PDA | ND | Cyanosis, RDS | Death at 16 h | 13   |
| 8, M             | 20 h, autopsy             | Dysplastic MV, PV stenosis, LV EFE | Cortical renal cysts, hydroureter/ nephrosis, microcephaly | Cyanosis, RD, cardiomegaly | Death at 20 h | 14   |
| 9, M             | 24 h, autopsy             | Hypoplastic LA and LV, LV EFE, ASD, PDA | Hemosiderosis of liver, minimal deposits in kidney and spleen | RD, cyanosis | Death at 24 h | 14   |
| 10, M            | 4 d, echo                | MV atresia, Ebstein malformation, TAPVR, PDA | Hemosiderosis of liver | Cyanosis | Death at 6 d | 14   |
| 11, F            | 18 wk, autopsy            | DORV, HLV, VSD, straddling of TV, MV atresia, absent PV | Hypoplastic nose, radial aplasia, absent thumbs, absent left index finger, intestinal malrotation, horseshoe kidney | Spontaneous abortion, severely macerated fetus | IUD at 18 wk | 15   |
| 12, M            | 18 wk, autopsy            | Absent PV, VSD, small MV, thickened LV | Cleft lip/palate, low set ears | Spontaneous abortion, severely macerated fetus | IUD at 18 wk | 15   |
| 13, F            | 18 wk, autopsy            | Complete AVSD, persistent LSVC, anomalous RSA | Nuchal bleb, edema, thymic hypoplasia, | Generalized edema | Abortion at 18 wk | 16   |

(Continued)
| Case no., gender | Diagnosis age and method | Other CHD | Noncardiac anomalies | Clinical presentation | Outcome | Ref. |
|------------------|--------------------------|-----------|----------------------|-----------------------|----------|------|
| 14, F            | 14 wk, autopsy           | DORV, VSD, absent PV, MV atresia, hypoplastic LA | Trisomy 13 on cytogenetics | –                     | Artificial abortion at 14 wk | 16   |
| 15, M            | 21 wk, autopsy           | DORV, VSD, absent PV | Cystic hygroma, single UA, absent thymus | Generalized edema | Artificial abortion at 21 wk | 16   |
| 16, ND           | 17 wk, fetal ECHO + autopsy | DORV, HLV, VSD, PDA | Hydranencephaly, hypoplastic left forearm, right radial aplasia | Hydrops, pericardial effusion, cardiomegaly | Termination at 18 wk | 17   |
| 17, M            | 9 h, 2D ECHO             | VSD, patent LSVC, absent MV, PDA, EFE of LV | High-arched palate, low set ears | Heart failure, cyanosis | Death at 20 h | 18   |
| 18, M            | 12 h, ECHO + cath        | HLH, EFE of LV, dysplastic MV | ND | Cyanosis, cardiomegaly | Norwood’s + BT shunt at 7 dol, death at 20 dol | 19   |
| 19, M            | ND, ECHO                 | Dysplastic LV, MV atresia, PDA | None | Mild cyanosis | Norwood’s at 12 dol, heart transplant at 1.5 mo | 3     |
| 20, M            | Fetal ECHO at 29 wk      | VSD, PDA, dysplastic PV | ND | Fetal hydrops | Death at 4 h | 20   |
| 21, M            | 2 d, ECHO                | MV atresia | ND | Cyanosis, Respiratory distress | Norwood’s at 9 d, BDG at 6 mo, Fontan’s at 6 y | 2.21 |
| 22, ND           | 31 wk, Fetal ECHO        | LV hypertrophy and EFE | Generalized lymphangiectasis, systemic air embolism | Fetal hydrops | Death at 2 h | 22   |
| 23, F            | 24 wk, Fetal ECHO        | DORV, large VSD, left SVC, AP window | Cystic hygroma, cerebral ventriculomegaly, microcephaly, clinodactyly, bilateral ear dysplasia, choanal atresia | Respiratory depression | Death at 2 d | 23   |
| 24, M            | 24 wk, Fetal ECHO        | HLH, MV atresia | Broad fingers | Cardiomegaly, cyanosis | Norwood’s and BDG at 3 mo, death at 10 mo | 1     |
| 25, M            | 23 wk, Fetal ECHO        | Dilated LV, EFE, hypoplastic MV | ND | Fetal hydrops | IUD at 28 w | 1     |

Abbreviations: AAV, absent aortic valve; AP, aorticopulmonary; ASD, atrial septal defect; AV, atrioventricular; AVSD, atrioventricular septal defect; BDG, bi-directional Glenn; CHD, congenital heart disease; d, day; dol, day of life; DORV, double outlet right ventricle; ECHO, echocardiogram; EFE, endocardial fibroelastosis; F, female; h, hour; HLH, hypoplastic left heart; HLV, hypoplastic left ventricle; IUD, intrauterine demise; L, left; LV, left ventricle; M, male, mo, months; MV, mitral valve; ND, not described; PDA, patent ductus arteriosus; PTH, parathyroid glands; PV, pulmonary valve; R, right; RA, right atrium; RD, respiratory distress; RSA, right subclavian artery; RV, right ventricle; SVC, superior vena cava; TAPVR, total anomalous pulmonary venous return; TV, tricuspid valve; UA, umbilical artery; wk, week; y, year.
reported cases to date. The rarity of this condition may be
due to the high rate of mortality leading to spontaneous
abortions. Even though it was initially thought to be an X-
linked recessive condition due to the first few reports being
males, that does not seem to be the case since a few female
cases have been reported.

Only two reported cases have survived beyond the first
few days of life. Harada et al\textsuperscript{2} described the first case of
successful palliative surgery in a patient with HLHS syn-
drome and AAV. At 9 days of life, the patient underwent
Norwood’s procedure with a Blalock-Taussig (BT) shunt.
The aortic valve was noted to be absent and left ventricular
cavity size was small with endocardial fibroelastosis. The
patient did well and underwent the bidirectional Glenn’s
procedure at the age of 6 months and the Fontan’s proce-
dure at the age of 6 years. The aortic annulus was not over-
sewn with the thought that coronary circulation would be
maintained owing to the markedly diminished size and
compliance of the LV. In addition, a balloon occlusion of
the left ventricular outflow tract showed no effect on the
coronary circulation.

The second case of AAV that survived beyond the first few
days of life was reported by Krasemann et al.\textsuperscript{3} Associated
cardiac defects included mitral atresia and a large noncon-
tractile hypoplastic LV. On day 12 of life the infant underwent
the Norwood’s procedure with additional surgical closure of
the aortic valve with the goal of preservation of coronary
perfusion. The postoperative course was complicated by
difficulty weaning from mechanical ventilation. An ortho-
tropic heart transplantation was successfully performed 4
weeks after the Norwood’s procedure. The postoperative
course was uneventful. The child was doing well at 5 months
of age when this case was reported.

The majority of reported cases of AAV were diagnosed
either on autopsy or on postnatal ECHO, with only a handful
of cases diagnosed on fetal ECHO (\textit{\textbf{Table 1}}). Fetal ECHO was
introduced as a diagnostic modality in the early 1970s and
has advanced significantly over the past few decades.\textsuperscript{4} It has
been shown to be a valuable tool in the optimization of
perinatal care.\textsuperscript{5,6} To date, no fetal interventions have been
attempted for the palliation of this condition.

Our case is one of the first few reported cases that were
diagnosed by fetal ECHO, the second case that underwent
successful palliative repair, and the third case that survived
beyond the first few days of life. To our knowledge, it is also
the first reported case in which the infant underwent suc-
cessful Norwood’s procedure with closure of the aortic valve
annulus. Closure of the aortic valve annulus would prevent
the development of coronary steal syndrome that may occur
if continued aortic regurgitation leads to increased left
ventricular size and compliance.

\section*{Conclusion}

AAV is rare and is usually associated with other congenital
cardiac anomalies, especially HLHS. It should be suspected in
the presence of aortic insufficiency on fetal ECHO. Palliative
repair with Norwood’s procedure with over-sewing of the
aortic annulus could potentially prevent coronary steal and
myocardial hypoperfusion in these patients.

\section*{Ethical Approval}

This article does not contain any studies with human
participants performed by any of the authors.

\section*{Informed Consent}

Informed consent was waived for this study since no
identifying information was included.

\section*{Conflict of Interest}

The authors declare that they have no conflicts of interest.

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