INTRODUCTION

Aortopulmonary window (APW) is a rare cardiac anomaly, accounting for 0.2%–0.6% of all congenital heart diseases.¹ The severe hemodynamic abnormalities caused by the continuous biphasic shunt significantly increase volume load and pulmonary over circulation, resulting in advanced left ventricular dysfunction and congestive heart failure.² The aortopulmonary septum defect should be closed immediately after diagnosis in all patients with APW.³ Therefore, prenatal diagnosis plays a very important role in the effective and timely treatment in these patients postpartum, thereby decreasing the mortality rate in neonates or infants. Echocardiography could provide valuable information for prenatal diagnosis and management of this condition. Thus, this study aimed to retrospectively analyze the prenatal APW echocardiographic manifestations, accompanied lesions, and gene expression and to compare the echocardiographic results to those of autopsy.

MATERIAL AND METHODS

2.1 Patient selection

We retrospectively reviewed the fetal echocardiographic database (n = 24 000) in our hospital between May 2012 and December 2017. Six pathology-confirmed APW cases were identified in our fetal echocardiographic database. On the three-vessel view, a communication between the pulmonary artery trunk and ascending aorta was noted above the two semilunar valves in all cases. The most frequent type of APW among the cases was type II, and all cases were associated with other cardiac anomalies. No pathogenic or suspected pathogenic copy number variation or insertion-deletions were detected in this series.

Conclusion: Prenatal diagnosis of APW is feasible, which is helpful during prenatal consultations, so that parents can make better decisions regarding postpartum treatment options and pregnancy outcomes.

KEYWORDS
aortopulmonary window, autopsy, fetal echocardiography, pregnancy outcome, prenatal diagnosis
with the agreement of the parents. The ethical committee of the hospital consented to the study, and the study was executed according to the ethical principles; informed consent was obtained from all the pregnant mothers.

The data recorded were as follows: maternal age, pregnancy screening (Down syndrome screening, noninvasive DNA or amniocentesis) results, gestational age (according to the last menstruation), reasons for undergoing fetal echocardiography, type and defect size of the APW, display view, associated cardiac abnormalities and pregnancy outcomes, autopsy results, and whole genome sequencing results.

### 2.2 | Echocardiographic equipment and image analysis methods

The ultrasound equipment used was Voluson E8, E10 (GE Healthcare, Zipf, AUSTRIA) equipped with 2D/3D probes (frequency range 4–8 MHz). Fetal echocardiography was performed by experienced ultrasound physicians. All cases were evaluated through a detailed fetal echocardiographic examination following standardized guidelines. General clinical data of pregnant women and fetal echocardiographic features were analyzed. Echocardiographic manifestations included the location and size of the aortopulmonary septal defect and associated cardiac abnormalities. APWs were classified into 3 types based on the aortopulmonary septal defect according to the criteria by Mori et al:

- APW defect at proximal (type I), distal (type II), and a combination of types I and II (type III).

### 2.3 | Genetics and pathological anatomy

In the case series, three pregnant women underwent Down syndrome screening, and three were screened through noninvasive DNA testing. Four cases underwent whole genome sequencing using fetal tissue.

All cases were examined through pathological anatomy.

### 3 | RESULTS

#### 3.1 | General clinical data

The incidence of APW was 0.25‰ in our database. All cases were referred to our department because they had an abnormal fetal heart pattern during a routine obstetric scan. The maternal age ranged from 22 to 39 years (mean 30.83 ± 2.41 years). The diagnostic gestation ages for APW were from 21.57 to 28.19 weeks (mean 24.29 ± 0.89 weeks).

#### 3.2 | Echocardiographic manifestation and associated cardiac anomalies of APW fetuses

The optimum view to diagnose APW is the 3-vessel view, wherein the communication between the ascending aorta and pulmonary artery can be identified. Five defects at the aortopulmonary septal distal part met the type II criterion (Figure 1A,B) and one defect at the aortopulmonary septal proximal part met the type I criterion (Figure 2A,B). The size of APW defects ranged from 3.4 to 7.4 mm (mean 4.74 ± 0.72 mm). The shunt direction between the aorta and pulmonary artery was bidirectional.

All cases were accompanied by other cardiac deformations prenatally, but not a significantly common associated anomaly. The detailed associated deformations were as follows: 2 cases of aortic coarctation, 2 cases of aortic origin of the right pulmonary artery, 3 cases of absence of arterial ducts, 2 cases of ventricular septal defect, 1 case of pulmonary artery stenosis, and 1 case of right ventricle double outlet.

#### 3.3 | Gene results

The risk associated with Down's screening and noninvasive DNA testing were low in these pregnant women. In this case series, 4 cases
had whole genome sequencing using fetal tissues without positive findings.

3.4 | Pathological results

There were two misdiagnosed cases: a case of interrupted aortic arch that was misdiagnosed as coarctation of the aorta and a case of an absence of the arterial duct that was misdiagnosed as absence of ductus arteriosus. The prenatal echocardiography diagnosis for APW is consistent with the results of the pathological anatomy.

The general clinical condition, main echocardiographic manifestations, chromosome, and autopsy outcome in all cases are listed in Table 1.

4 | DISCUSSION

Failure of the aortopulmonary septum to divide the arterial trunk into the ascending aorta and the pulmonary artery during development can lead to APWs. APW includes a communication between the ascending aorta and the pulmonary artery and the normal anatomy of the aortic and pulmonary valves. Berry supplemented the type II cases based on the Mori category method. Berry syndrome occurs when a type II APW is associated with the aortic origin of the right pulmonary artery, interrupted aortic arch or coarctation of the aorta, and intact ventricular septum.

Aortopulmonary window can occur as an isolated anomaly or accompanied by other lesions. In this case series, five cases were classified as Type II, one case was classified as Type I, and all cases

### TABLE 1 General clinical condition, main echocardiographic manifestations, and autopsy outcome in all APW cases

<table>
<thead>
<tr>
<th>Case</th>
<th>Age</th>
<th>G</th>
<th>P</th>
<th>GA</th>
<th>Prenatal diagnosis</th>
<th>Chromosome</th>
<th>Autopsy outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>32</td>
<td>3</td>
<td>1</td>
<td>24.43</td>
<td>APW (II), VSD, COA</td>
<td>Normal</td>
<td>APW (II), VSD, COA</td>
</tr>
<tr>
<td>2</td>
<td>35</td>
<td>5</td>
<td>1</td>
<td>24.43</td>
<td>APW (I), absence arterial duct</td>
<td>Normal</td>
<td>APW (I), absence arterial duct</td>
</tr>
<tr>
<td>3</td>
<td>29</td>
<td>1</td>
<td>0</td>
<td>23.43</td>
<td>APW (II), COA, AORPA</td>
<td>Normal</td>
<td>APW (II), IAA, AORPA</td>
</tr>
<tr>
<td>4</td>
<td>39</td>
<td>5</td>
<td>1</td>
<td>21.57</td>
<td>APW (II) absence arterial duct</td>
<td>None</td>
<td>APW (II)</td>
</tr>
<tr>
<td>5</td>
<td>23</td>
<td>1</td>
<td>0</td>
<td>23.71</td>
<td>APW (II), AORPA</td>
<td>Normal</td>
<td>APW (II), AORPA</td>
</tr>
<tr>
<td>6</td>
<td>22</td>
<td>1</td>
<td>0</td>
<td>28.29</td>
<td>APW (II) DORV, PA, absence arterial duct VSD</td>
<td>None</td>
<td>APW (II) DORV, PS, absence arterial duct VSD</td>
</tr>
</tbody>
</table>

AORPA = aortic origin of the right pulmonary artery; COA = coarctation of the aorta; DORV = double outlet right ventricle; IAA = interrupted of aortic arch; PA = pulmonary atresia; PS = pulmonary stenosis; VSD = ventricular septal defects.
were accompanied by other cardiac anomalies during the prenatal period. The associated cardiac anomalies include ventricular septal defects (Figure 3A–C), interrupted aortic arch, coarctation of the aorta, aortic origin of the right pulmonary artery (Figure 4A–C), double outlet right ventricle, and pulmonary stenosis. One case met the Berry syndrome criteria. The results were a little different from previous publications, which reported that type I was...
the most common form, and the most common complication was interrupted aortic arch (5 of 10 [50%]). The discrepancy may be related to the small sample size.

Prenatal echocardiography can achieve the ideal 3-vessel view without disturbance from lung gas. The view is similar to the postnatal parasternal short view, but without gas interference. To avoid dropout of false echoes, the probe beam is adjusted perpendicular to the fetal pulmonary artery and ascending aorta. The view is the optimal window to scan the size and position of the APW. At this view, the aortopulmonary septal discontinuation and the shunt between the ascending aorta and pulmonary artery were observed. In these cases, the shunt direction between the ascending aorta and pulmonary artery is bidirectional, which may be related to the hemodynamic characteristics of the fetal phase and complicated with cardiac malformation. However, in some cases with an abnormal artery, the standard 3-vessel view is difficult to achieve.

Compared with autopsy, prenatal echocardiography has high specificity and sensitivity in diagnosing APW. However, when APWs are associated with other cardiac malformations, the accurate diagnosis of APW and all its associated abnormalities can be challenging. In our case series, two cases were misdiagnosed: a case of IAA that was misdiagnosed as COA and a case of arterial duct that was misdiagnosed as absence of ductus arteriosus.

Genome sequencing performed in four patients revealed no chromosomal abnormality in all the four patients, which is similar to the findings of previous studies. These scholars believe that APW is not associated with chromosomal anomalies.

The treatment effect is related to the cardiac malformations and pulmonary hypertension. Talwar et al have investigated postoperative hospital mortality depending on the associated lesions by dividing the APW cases into simple and complex groups. The hospital mortality in the simple and complex groups was 6.97% and 21.05%, respectively. Some experts have reported that APW without other associated lesions can be closed by a percutaneous or transcatheter device, with better outcomes. Therefore, if treated promptly, a better prognosis can be achieved in most patients with APW.

However, in the case series, the pregnant women chose to terminate their pregnancy. It is very important therefore to increase the understanding of APW and to provide accurate guidance for pregnant women during prenatal consultations.

The limitations of this study include a small sample size and absence of postnatal follow-up and prognosis. In future studies, attention should be focused on prognosis at postpartum and the therapeutic effect of different therapies on fetuses with APW.

## 5 | CONCLUSION

Prenatal diagnosis of APW by fetal echocardiography is feasible, which can provide information for an effective postpartum treatment plan, but the accurate diagnosis of all associated abnormalities can be challenging. It is necessary to provide more information for the parents of fetuses with APW during prenatal consultations, so that they can make better decisions for pregnancy outcome and can receive timely postpartum interventions.

## AUTHOR CONTRIBUTIONS

Shaomei Yu, MD, performed data collection, writing article. Jiancheng Han, MD, performed data collection, critical revision of article. Shuang Gao, MD, Xiaowei Liu, MD, Xiaoyan Gu, MD, Ye Zhang, MD, and Lin Sun, MD performed data collection. Yihua He, MD, performed critical revision of article data collection, approval of article.

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## REFERENCES


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