Fetal Anatomy: The Aortic Valve in Fetal Aortic Valve Diseases

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5.1 Introduction

As in the adults, also in the fetus, the pathology of aortic valve diseases can manifest at subvalvular, valvular, or supravalvular level [1–4]. Fetal supravalvular aortic stenosis is very rare, while subvalvular stenosis is encountered more often in the setting of complex congenital defect, like posterior malalignment of conal septum, subaortic conus, or mitral valve tissue [5]. The most common fetal aortic valve diseases are represented by atresia or valvular stenosis.

5.2 Morphological features

Aortic valvular stenosis is often characterized by restricted cusps excursion and post-stenotic dilatation of the ascending aorta. In aortic atresia an imperforate membrane is guarding the aortic severe hypoplastic annulus and ascending aorta (Fig. 5.1).

Congenital aortic stenosis is a very rare condition falling in the spectrum of congenital left heart obstructions, encompassing a wide range of morphological features, from tricuspid aortic valve dysplasia or asymptomatic bicuspid aortic valve to unicuspid, severely stenotic valve to complete atresia and can be part of the hypoplastic left heart syndrome [1, 2, 6–9].

As in critical pulmonary valve stenosis, also in critical aortic valve stenosis, the blood flow and pressure during gestation impact on the remodeling cardiac process

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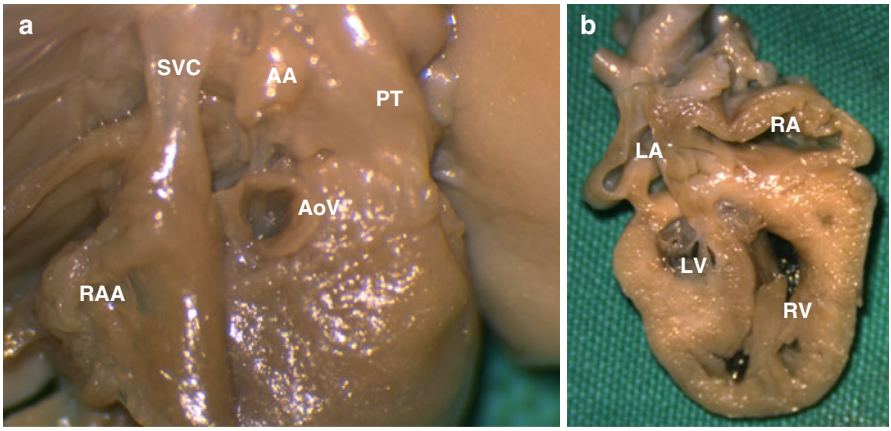

Fig. 5.1 Aortic valve atresia with imperforate valve. (**a**) View of the imperforate valve from above, after sectioning of the ascending aorta. Three well-formed commissures are still identifiable. (**b**) Four-chamber cut of the heart, showing the well-formed right ventricle reaching the apex and the dilated right atrium. Note the severe hypoplasia of the left ventricle, with hypertrophic free wall and minute mitral valve. *AA* ascending aorta, *AoV* aortic valve, *LA* left atrium, *RA* right atrium, *RAA* right atrial appendage, *RV* right ventricle, *SVC* superior vena cava, *PT* pulmonary trunk

and can lead to hypoplastic left heart syndrome (HLHS) due to impairment in left ventricle and aortic arch growth [10, 11].

HLHS with mitral and aortic stenosis manifests with severe aortic stenosis and left ventricular remodeling of different degree: at midtrimester of gestation, with hypoplasia, or dilatation, if associated to mitral valve incompetence; later in the gestational period, critical aortic stenosis can occur with a borderline left ventricle or even a relatively normal ventricle. In some setting the dimensions of the left ventricle remain unchanged throughout the gestational period [12–14].

Critical aortic valve stenosis can be due to an anomaly in the number of cusps, namely, unicuspid, bicuspid, or even quadricuspid, and/or to dysplasia of the cusps. The structure of the cusp is composed by the fibrosa covered by the ventricularis and the arterialis and by the spongiosa located on the ventricular side between the fibrosa and the ventricularis. In the setting of dysplasia, the structure is altered with loss of fibrosa integrity and mucoid degeneration and nodular thickening. The dysplastic cusps are usually thicker and rigid than the normal ones.

In the setting of unicuspid valve, there are one commissure with an eccentric intrinsically stenotic commissural orifice, only one well-formed interleaflet triangle, and a small aortic annulus and dysplastic cusp with myxoid nodular excrescences usually located on the ventricular aspect of the valve (Figs. 5.2 and 5.3). Two raphes can be identified as remnants of the commissures indicating lack of cusp separation or fusion. Bicuspid aortic valve is usually non-stenotic, and stenosis is usually present when there is associated dysplasia of the cusps (Fig. 5.4). Even tricuspid valve can be stenotic due to cusp dysplasia. The ascending aorta and aortic arch can be hypoplastic to a different extent. The left cavities can present with different patterns:

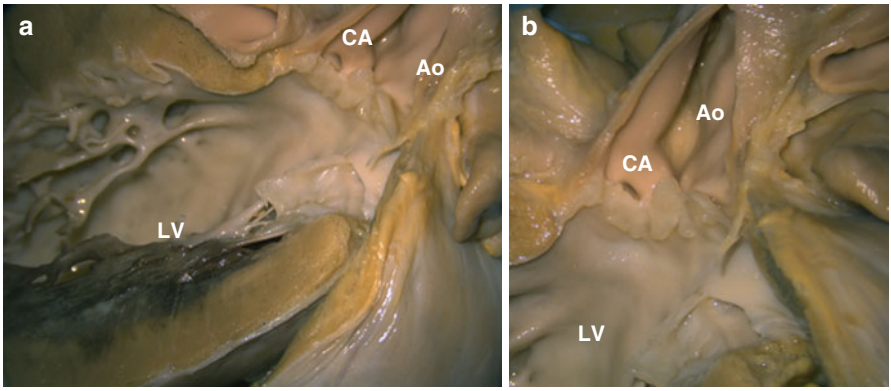

Fig. 5.2 Critical aortic valve stenosis with dysplastic valve. (**a**) View of the left ventricle and the aorta: note the unicuspid valve with severe dysplastic thickening and one identifiable commissure and the left ventricle with whitish thickened endocardium suggestive of endocardial fibroelastosis. (**b**) Close-up of the same specimen showing the hypoplasia of the annulus and the origin of the right coronary artery. *Ao* aorta, *CA* coronary artery, *LV* left ventricle

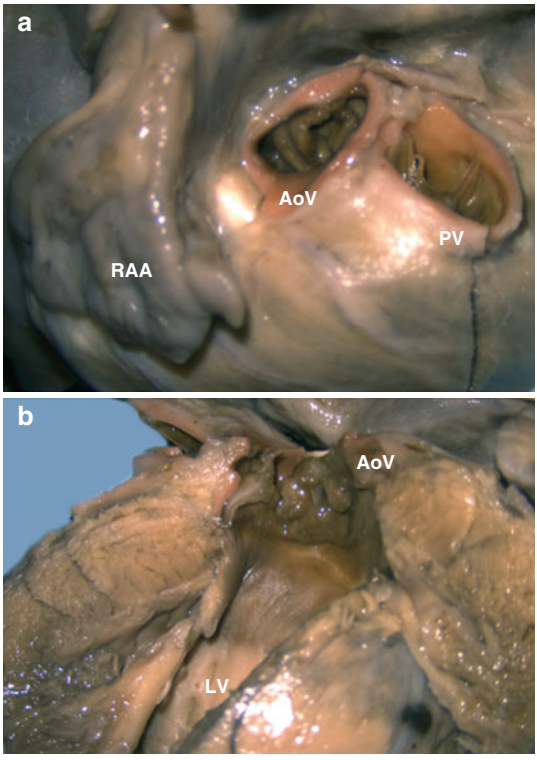

Fig. 5.3 Critical aortic valve stenosis with unicuspid dysplastic valve. (**a**) Right lateral view of the great arteries and of the right atrial appendage: the aortic valve present hypoplasia of the anulus and an eccentric orifice. (**b**) View from the left outflow tract showing the dysplastic cusp with nodular thickening. *AoV* aortic valve, *PV* pulmonary valve, *RAA* right atrial appendage

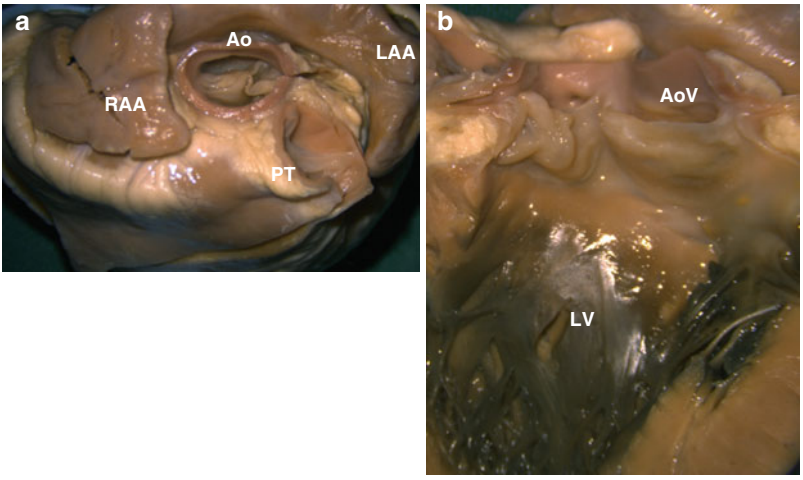

Fig. 5.4 Aortic valve stenosis with bicuspid valve. (**a**) View from the above of the great arteries and of the right and left atrial appendages two well formed interleaflets triangles and one aborted triangle in relation to the raphe are present. (**b**) View from the left outflow tract showing the two cusps, the anterior one with the raphe and dysplasia of the cusp. *AAo* ascending aorta, *LV* left ventricle

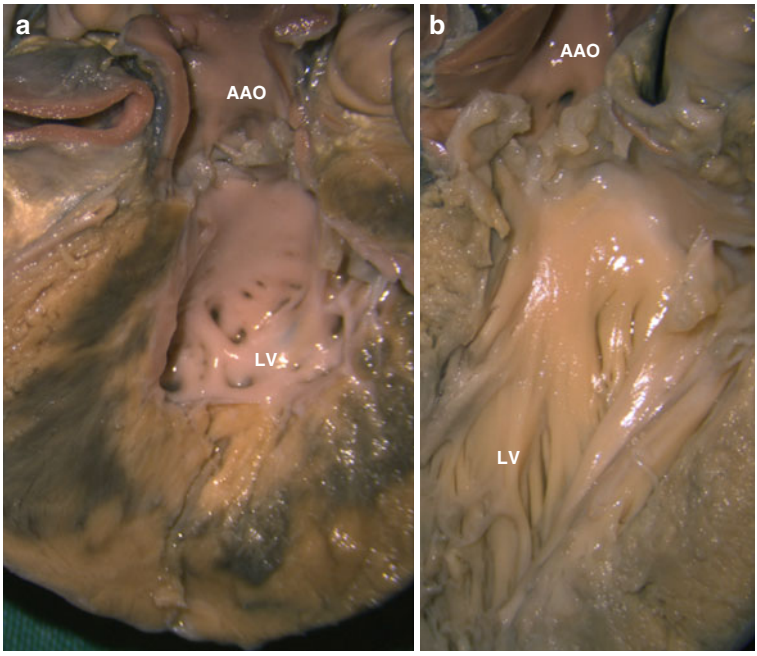

Fig. 5.5 Critical aortic valve stenosis with unicuspid valve. (**a**) View from the left outflow tract showing the dysplasia of the cusp and the small left ventricle with hypertrophy of the parietal wall and severe diffuse endocardial fibroelastosis. (**b**) View from the left outflow tract showing the dysplasia of the cusp and a more developed left ventricle and less severe endocardial fibroelastosis confined to the lateral wall. *RAA* right atrial appendage, *LAA* left atrial appendage, *Ao* aorta, *PT* pulmonary trunk, *AoV* aortic valve, *LV* left ventricle

Critical aortic stenosis and HLHS can be associated to severely restricted or intact atrial septum. With left heart obstructive lesions, there is left atrial hypertension leading to severe pulmonary vein dilatation and altered blood flow, which can produce dilated lymphatics and arterialization of the pulmonary veins.

Valve stenosis is due, in the majority of cases, not only to the fusion of the cusps with the presence of rudimentary commissures but also (sometimes mainly) to irregular cups thickening. These excrescences protrude into valve orifice and hamper its opening. Endocardial fibroelastosis is usually associated with critical aortic stenosis and can be focal, involving the papillary muscle or the septum, or can be diffuse to all the ventricular cavity with, at macroscopic evaluation, whitish appearance of the endocardium and severe thickening due to fibroelastic fiber endocardial deposition [15] (Fig. 5.5). There is no association between the severity of the endocardial fibroelastosis and size of the aortic valve and leaflet.

Conclusions

Aortic valve disease can present with a wide spectrum of abnormalities, at subvalvular, valvular or supravalvular level. Heterogeneity affects the number of cusps, grade of dysplasia of the cusp, and is associated with aortic annular hypoplasia. This is directed related to the number of cusps. Critical aortic valve stenosis or atresia is associated to hypoplasia of the left ventricle, mitral valve involvement and usually endocardial fibroelastosis. If mitral valve incompetence is present the left ventricle can be dilated with thin parietal wall and giant left atrium.

References

1. Frescura C, Ho SY, Thiene G. La collezione anatomica di cardiopatie congenite dell'Università di Padova. Cleup, Padova 1996.
2. Anderson RH, Devine WA, Ho SY, Smith A, McKay R. The myth of the aortic annulus: the anatomy of the subaortic outflow tract. Ann Thorac Surg. 1991;52:640–6. J Heart Valve Dis. 5 Suppl 3:S272-5.
3. Ho SY. Structure and anatomy of the aortic root. Eur J Echocardiogr. 2009;10:i3–10.
4. Sievers HH, Hemmer W, Beyersdorf F, Moritz A, Moosdorf R, Lichtenberg A, Misfeld M, Charitos EI, Working Group for Aortic Valve Surgery of German Society of Thoracic and Cardiovascular Surgery. The everyday used nomenclature of the aortic root components: the tower of Babel? (2012). Eur J Cardiothorac Surg. 2012;41:478–82.
5. Yamamoto Yand Hornberger LK. Progression of outflow tract obstruction in the fetus. Early Hum Dev. 2012;88:279–85.
6. Angelini A, Ho SY, Anderson RH, Devine WA, Zuberbuhler JR, Becker AE, Davies MJ. The morphology of the normal aortic valve as compared with the aortic valve having two leaflets. J Thorac Cardiovasc Surg. 1989;98:362–7.
7. Duran AC, Frescura C, Sans-Coma V, Angelini A, Basso C, Thiene G. Bicuspid aortic valves in hearts with other congenital heart disease. J Heart Valve Dis. 1995;4:581–90.
8. Frescura C, Thiene G. Small aortic root in neonates. J Heart Valve Dis. 1996;Suppl 3:S272–5.
9. Maizza AF, Ho SY, Anderson RH. Obstruction of the left ventricular outflow tract: anatomical observations and surgical implications. J Heart Valve Dis. 1993;2:66–79.
10. Arzt W, Wertaschnigg D, Veit I, Klement F, Gitter R, Tulzer G. Intrauterine aortic valvuloplasty in fetuses with critical aortic stenosis: experience and results of 24 procedures. Ultrasound Obstet Gynecol. 2011;37:689–95.

11. Freud LR, McElhinney DB, Marshall AC, Marx GR, Friedman KG, del Nido PJ, Emani SM, Lafranchi T, Silva V, Wilkins-Haug LE, Benson CB, Lock JE, Tworetzky W. Fetal aortic valvuloplasty for evolving hypoplastic left heart syndrome: postnatal outcomes of the first 100 patients. Circulation. 2014;130:638–45.
12. Marantz P, Grinenco S. Fetal intervention for critical aortic stenosis: advances, research and postnatal follow-up. Curr Opin Cardiol. 2015;30:89–94.
13. Maskatia SA, Ing FF, Justin H, Crystal MA, Mullins CE, Mattamal RJ, O'Brian Smith E, Petit CJ. Twenty-five year experience with balloon aortic valvuloplasty for congenital aortic stenosis. Am J Cardiol. 2011;108:1024–8.
14. McKay R, Smith A, Leung MP, Arnold R, Anderson RH. Morphology of the ventriculoaortic junction in critical aortic stenosis. Implication for hemodynamic function and clinical management. J Thorac Cardiovasc Surg. 1992;104:434–2.
15. McElhinney DB, Vogel M, Benson CB, Marshall AC, Wilkins-Haug LE, Silva V, Tworetzky W. Assessment of left ventricular endocardial fibroelastosis in fetuses with aortic stenosis and evolving hypoplastic left heart syndrome. Am J Cardiol. 2010;106:1792–7.