Aplasia of Semilunar Valve Leaflets: Two Case Reports and Developmental Aspects

Nico G. Hartwig, Christl Vermeij-Keers, Henrike E. De Vries, and Adriana C. Gittenberger-De Groot

Department of Anatomy and Embryology, University of Leiden, Leiden, The Netherlands

SUMMARY. Two fetuses of approximately 11 weeks development with aplasia of the pulmonary as well as the aortic valve leaflets are reported. Both cases showed additional cardiac malformations. Case 1, with all leaflets missing, also had double-outlet right ventricle, hypoplastic left ventricle, large ventricular septal defect, straddling tricuspid valve, and atretic mitral valve. Case 2, with only one hypoplastic aortic valve leaflet, showed hypoplasia of the mitral valve and the left ventricle, and a subaortic ventricular septal defect.

The observations made and data in the literature suggest that aplasia of semilunar valve leaflets reflects an underdevelopment of the endocardial cushion swellings at the ventriculoarterial junction, rather than resulting primarily from a malseptation of the cardiac outflow tract.

KEY WORDS: Congenital heart defect — Embryology — Aortic valve — Pulmonary valve — Semilunar valve leaflet asplasia

Absence or aplasia of semilunar valve leaflets is a rare congenital cardiac malformation [8]. It may involve one or more leaflets, but is restricted to one semilunar valve at a time [3, 6, 8, 12]. However, we recently encountered two fetuses with aplasia of semilunar valve leaflets in both the pulmonary and the aortic valves. In describing these cases, particular attention is paid to certain developmental aspects of the semilunar valves.

Case Reports

Case 1

Patient 1 was a 24-year-old gravida 1, para 0. At 18 weeks' amenorrhea her pregnancy terminated with a spontaneous abortion. The expelled fetus (Fig. 1) had a crown-rump length (CRL) of 62 mm (in conformity with 13 weeks' amenorrhea) and was severely macerated. Macroscopically, the following abnormalities were observed: a hypoplastic nose, bilateral radial aplasia, bilateral absence of the thumb, absence of the left index finger, a small left ventricle, malrotation of the intestines, and a horseshoe kidney.

As the heart was too small for further macroscopic examination, it was serially sectioned at 10 μm and microscopically investigated. In both the aorta and the pulmonary trunk no semilunar valve leaflets were present (Fig. 2A and B). Where normal leaflet insertion should occur there was only a small ridge of mesenchymal tissue. Above this level the coronary arteries originated normally from the aorta, taking their respective course in the atrioventricular sulcus. Both great vessels showed a thickening of the tunica media, the pulmonary trunk from its origin into the ductus arteriosus, and the aorta in its ascending part. For the greater part, the aorta was situated above the right ventricle and there was no fibrous connection between the orifices of the aortic and mitral valves, hence the diagnosis "double-outlet right ventricle." Furthermore, the heart showed an enlarged right ventricle, hypoplasia of the left ventricle, a ventricular septal defect, wide straddling of the tricuspid valve, and an atretic mitral valve.

Case 2

Patient 2 was a 27-year-old gravida 1, para 0. During a routine obstetrical check-up at 18 weeks' amenorrhea, intrauterine death was noted. Ultrasound revealed a dead fetus which corresponded to 12 weeks' amenorrhea. Labor was induced by prostaglandin infusion. The fetus (Fig. 3) with a CRL of 64 mm was severely macerated. Macroscopically, a cleft lip and palate on the right, low set ears, and incomplete fusion of the scrotal swellings were observed.

The heart, serially sectioned, revealed several structural defects. In the enlarged pulmonary trunk there were no semilunar valve leaflets; the supposed site of insertion was marked by a small ridge of mesenchymal tissue. The aortic valve had only one hypoplastic leaflet (the left one), the two other leaflets were absent (Fig. 4A and B). Both coronary arteries originated from the
Fig. 1. External appearance of case 1.

Fig. 2. (A) Microscopic section through the heart of case 1 at the arterial pole, just below the aortic orifice (a). In the pulmonary orifice (p), no valve leaflets are present. The arrows indicate the coronary arteries. The ventricle shown in this section represents the right ventricle (rv). Original magnification, ×16. (B) Microscopic section through the aortic orifice (a). No valve leaflets are present. Original magnification, ×16.

Fig. 3. External appearance of case 2.

Fig. 4. (A) Microscopic section through the heart of case 2 at the pulmonary orifice (p). No valve leaflets are present. Part of the right ventricle (rv) is shown in this section. Original magnification, ×16. (B) Microscopic section through the aortic orifice (a). The aortic valve (a) shows one hypoplastic valve leaflet. The pulmonary trunk (p) is enlarged. Original magnification, ×16.

Further, a subaortic ventricular septal defect, a relatively small mitral valve, and a thickened left ventricular wall were noted.

Discussion

Although malformations of the semilunar valves are the most frequent congenital heart malformations [5], absence or aplasia of leaflets is rarely seen. It may involve one or more leaflets [3, 8, 11], but is restricted to one semilunar valve at a time [3, 6, 12], the pulmonary valve being affected most [12]. The two fetuses reported here are the first to have aplasia of both pulmonary and aortic valve leaflets. Case 1 has been briefly mentioned before as an example of abnormal valve development [2]. How-