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Ology Press

Proceeding

Advances in Cardiology and Cardiovascular Disorders

Familial bicuspid aortic valve in pregnancy - a case report

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Received: November 10, 2017 | **Published:** November 14, 2017

Bicuspid aortic valve (BAV) is the most common adult congenital cardiac malformation, occurring in 0.5–2% of the population. The malformation involves both the valve and the aorta, and patients require lifelong surveillance. The genetic determinants of BAV and its complications remain largely undefined. Just as BAV phenotypes are highly variable; the genetic aetiologies of BAV are equally diverse and vary from complex inheritance in families to sporadic cases without any evidence of inheritance.

We report here a 26-year-old primigravida, a known case of BAV diagnosed in infancy, had undergone a balloon aortic valvotomy at 4 years and subsequent Ross procedure at 12 years of age. In view of pulmonary regurgitation, she underwent a transcatheter pulmonary valve implantation two years back. During her antenatal evaluation and follow up, fetal echocardiogram evaluation revealed a hypoplastic left ventricle with flow reversal in aortic arch, hypoplastic ascending aorta with probable aortic atresia. First trimester double marker screening was suggestive of low risk for fetal aneuploidies. Regular fetal monitoring done showed normal interval growth of fetus. Her cardiac evaluation showed good biventricular function with mild residual right ventricular obstruction. Pedigree analysis revealed her father was diagnosed to have a BAV with severe aortic stenosis. He underwent Ross procedure at 42 years of age. Her grandfather also died of cardiac disease at the age of 47.

BAV occasionally demonstrates complex inheritance in large families without syndromic features. Autosomal-dominant transmission of BAV was observed in some 3-generation pedigrees, but no single-gene model clearly explains BAV inheritance. The prevalence of BAV stands nearly 10-fold higher in primary relatives of patients with BAV than in the general population. Although the genetic links remain unknown, BAV is also seen with some congenital heart disorders involving the left ventricular outflow tract (LVOT), such as hypoplastic left heart syndrome (HLHS), coarctation of the aorta (CoA), and some ventricular septal defects. HLHS and CoA are significantly enriched among primary relatives of patients with BAV, and according to linkage analysis, mutations in the same genes probably caused some cases of HLHS and BAV. One possible interpretation of these observations is that LVOT morphology and obstruction may also influence aortic valve development because of alterations in blood flow or shear stress.

Whole exome sequencing (WES) studies are now underway to identify the definitive cause of this disease. Pre pregnancy evaluation of cardiac function and vigilant antepartum surveillance are important components of a safe pregnancy for women with BAV. In cases of familial BAV, as there is increased chance of HLHS and CoA, antenatal diagnosis of fetal cardiac anomalies will help to improve pregnancy outcomes.