

# Bicuspid aortic valve

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## Abstract

The usual arrangement of the aortic valve is for it to have three leaflets; these are termed the right and left coronary leaflets and the non-coronary leaflet. Bicuspid aortic valve (BAV) describes a valve with two leaflets. It can be detected using cross-sectional and Doppler echocardiography. Post mortem and echocardiographic studies give a prevalence of 0.5% to 2.0% with an excess of males. Bicuspid aortic valves are often associated with other cardiovascular malformations, coarctation of the aorta and aortic dilatation. The aorta is often dilated even with a normal functioning valve. Bicuspid aortic valve is associated with a number of complications; aortic stenosis, aortic regurgitation, aortic dissection, and infective endocarditis.

Individuals with BAV should be carefully monitored for evidence of aortic dilatation and valvular dysfunction.

There are reports of the familial nature of BAV that are consistent with autosomal dominant inheritance with reduced penetrance especially in females. First-degree relatives should be offered screening for BAV and the complications.

## Key Words

Bicuspid aortic valve, coarctation of the aorta, aortic dilatation, aortic dissection, endocarditis, autosomal dominant

## Disease Name

Bicuspid Aortic valve

Bicommissural Aortic valve.

## Definition/diagnostic criteria

The usual three-leaflet arrangement of the aortic valve is replaced by two leaflets.

## Etiology

BAV is a malformation of the aortic valve. Two of the three leaflets are formed and there is evidence of a raphe between the conjoined leaflets. The commonest arrangement is for

the two coronary leaflets to be joined (Sabet, 1999).

It is seen in a number of conditions, e.g. [Turner's Syndrome](#) (Slater, 1982) (Miller, 1983) [retinoic acid embryopathy](#) (Lammer, 1985).

The cause of isolated BAV is unknown (Fedak, 2002).

## Clinical description

There are no pathognomic clinical findings; there may be aortic ejection click and an ejection systolic murmur (Ward, 2000).

BAV is found in association with other congenital heart defects (Fernandes, 2004) including [coarctation of the aorta](#), (Abbott,

1928) [interruption of the aorta](#) and [ventricular septal defects](#) (Duran, 1995).

The complications of a bicuspid aortic valve are:

Aortic stenosis that may be present in newborns (Moller, 1966). Ward (Ward, 2000) suggested that 50% of adults with aortic stenosis have a bicuspid aortic valve. This may reflect a propensity to premature fibrosis, and calcification.

Aortic regurgitation may occur in isolation (Roberts, 1981). but also in association with aortic dilatation, coarctation of the aorta or infective endocarditis.

McKusick (McKusick, 1957) reported an association of BAV with Erdheim's cystic medial necrosis. This suggests that dilation of the aorta is a manifestation of an abnormal aortic media. Dilatation of the aorta may occur in association with a normally functioning bicuspid valve (Dore, 2003, Gurvitz, 2004).

Aortic dissection does occur in association with BAV (McKusick, 1957). The cause of this is unknown but is likely to be related to an intrinsic weakness of the aortic wall. (Lindsay, 1988)

It is likely that BAV, coarctation of the aorta and aortic dilatation represent a developmental defect of the arterial tree (Lindsay, 1988, Warnes, 2003).

### Diagnostic Methods

The introduction of cross sectional and Doppler echocardiography has improved detection. Cross sectional echocardiography can visualize the two cusps and two commissures of the bicuspid aortic valve with a parasternal short axis view (Brandenburg, 1983)

BAV may be detected at post mortem with pathological examination of surgically removed valves (Sabet, 1999).

### Epidemiology

Basso *et al* (Basso, 2004) screened 817 healthy 10 year olds for BAV, they found it in 0.5% with an excess of males (75% male). This figure is less than the commonly quoted figure of 1-2%, this data derives from post mortem studies.

There have been several reports of familial cases. However with a prevalence of 1% it would not be surprising to see familial cases. These reports suggested autosomal dominant inheritance. (Clementi, 1996, Huntington, 1997). Cripe *et al* undertook a study that showed a high heredity for BAV (Cripe, 2004).

The proportion of individuals with BAV who will have a significant complication is unknown. Wood estimated that it was between one third

and the majority of individuals will develop complications (Wood, 2000).

### Genetic Counselling

As BAV is a familial disease and has a high risk of complications, first-degree relatives should be offered screening by echocardiography.

### Management

There are a number of complications that individuals with BAV are at risk of, though the exact risk is not known.

Endocarditis was described by Osler (Osler 1886). Individuals should be given advice about antibiotic prophylaxis before surgical treatment.

Aortic valve stenosis and regurgitation and aortic root dilatation may develop and so individuals should be offered regular follow up which should include checking for hypertension and imaging of the ascending aorta by echocardiography. The value of prophylactic  $\beta$ -blockers as used in [Marfan syndrome](#) is unknown.

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