Background Bicuspid aortic valve (BAV) is the commonest valvular congenital heart defect, affecting around 1% of the population. BAV shows strong familial clustering and is thought to be inherited as an oligogenic, autosomal dominant trait with incomplete penetrance. Guidelines recommend cascade screening of first degree relatives of patients with BAV using echocardiography but the effectiveness of this approach is unknown. Incomplete penetrance can negatively influence screening programs leading to non-detection of silent carriers.
Aim To assess the prevalence of incomplete penetrance of BAV among families with the familial form of the disease.
Methods Consecutive patients with a diagnosis of BAV presenting to Glenfield Hospital, Leicester were recruited. First degree relatives were screened for the presence of BAV using transthoracic echocardiography. In families with at least two affected individuals, screening was extended to second-degree relatives.
Results 425 participants were recruited to the study (298 with BAV, 127 unaffected relatives). 16 multigenerational pedigrees with multiple affected subjects were identified. Incomplete penetrance was observed in three of sixteen pedigrees (19%), meaning that echocardiographic screening demonstrated the absence of BAV in a particular generation and the presence of BAV in the subsequent generation of the same lineage
Conclusions Incomplete penetrance is common in pedigrees with familial forms of BAV. This phenomenon may reduce the effectiveness of cascade screening for BAV using echocardiography alone as it fails to identify silent carriers. A better understanding of the genetics of familial BAV might lead to improvements in the effectiveness of cascade screening programmes for the condition.

13 TRANSCATHETER AORTIC VALVE IMPLANTATION IN ADULT CONGENITAL HEART DISEASE – SINGLE CENTRE EXPERIENCE

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Background Transcatheter aortic valve implantation (TAVI) is reserved for inoperable aortic stenosis. The use of TAVI for AS in the adult congenital heart disease (ACHD) population is not well described. We aim to assess the effectiveness of TAVI for treatment of aortic stenosis in the ACHD population.
Methods A retrospective review of cardiac catheterisation reports and medical notes of all patients that underwent TAVI from January 2008 to August 2016. 4 ACHD patients were identified from 329 TAVI procedures performed at the Bristol Heart Institute. All patients were declined for surgery by surgical team in the multi-disciplinary team meeting. Patients received either a Core Valve Evolut R or Edwards Sapien TAVI valve based on their aortic valve anatomy.
Results All 4 patients (3 Male, 1 Female) had different underlying congenital anatomy (calcified aortic valve homograft, congenitally-corrected transposition of great arteries, bicuspid aortic valve with coarctation aneurysm, and atriopulmonary Fontan with bicuspid aortic valve respectively). This is shown in Table 1. Median age was 66.7 (range 29–81). Mean aortic annulus size was 27 mm (range 24 mm – 30 mm), mean echo pre-procedural peak gradient was 66 mmHg (range 47 mmHg – 85 mmHg), and mean echo post-procedural peak gradient was 22 mmHg (range 21 mmHg – 23 mmHg). 2 Core Valve Evolut R (29 mm and 31 mm) and 2 Edwards Sapien S3 valve (23 mm) were implanted. Median stay in hospital was 13.0 days (range 6–28). 1 patient required a pacemaker post TAVI. No severe post-TAVI paravalvular leak. All patients had reduction in NYHA class post TAVI. 1 patient died at 4 months post-TAVI from recurrent aortic valve endocarditis and 1 patient died at 7 months post-TAVI unrelated to the procedure.
Conclusion TAVI is potentially an evolving therapy for inoperable aortic stenosis in ACHD patients with good symptomatic relief. Further experience with the use of TAVI in the ACHD patients is required to assess long-term outcomes in unique group of patients.

Abstract 13 Table 1

	Age Gender	Congenital Anatomy	NYHA Class	Valve	Peak echo gradient (mmHg)	Pacemaker intra-procedural	Length of stay (days)	Clinical Status
Patient #1	53 Female	Tricuspid atresia Atrio-pulmonary Fontan Bicuspid aortic valve	Pre 3 Post 1	Medtronic Core Valve Evolut R 29 mm	Pre 85 Post 21	Yes	10	Alive
Patient #2	80 Male	Coarctation Bicuspid aortic valve	Pre 3 Post 1	Edwards Sapien S3 23 mm	Pre 83 Post 23	No	6	Alive
Patient #3	81 Male	Double discordance TGA Calcified trileaflet aortic valve	Pre 3 Post 1	Medtronic Core Valve 31 mm	Pre 49 Post 23	No	16	Died at 7 months post TAVI at DGH due to presumed pneumonia
Patient #4	29 Male	Coarctation Calcified aortic homograft	Pre 4 Post 2	Edwards Sapien XT 23 mm	Pre 47 Post 21	No	28	Died at 4 months post TAVI due to endocarditis