Results We identified 8 patients (6 female) with mean age 26.3±6.2 years. Mean follow-up was 4.1 years (range, 18 months to 12 years). All patients have had surgical repair; one patient required concomitant mitral valve repair. Mean age at operation was 9.7 years (range, 3 months to 34 years old); 3 patients (38%) were operated as adults. At last clinic review, one patient had NYHA III symptoms, the rest were well (NYHA I). Mean peak oxygen consumption on cardiopulmonary exercise testing was 36±2.4 ml/kg/min (range 33.4 to 38.8 ml/kg/min, mean 93%±15.9% predicted, range 78% to 112%). All patients were in sinus rhythm and no ischaemia or arrhythmias were identified. 7 patients (88%) had good left ventricular function (mean EF 61%); 1 patient had mildly impaired function (EF 50%) due to an apical transmural infarction. Moderate mitral regurgitation was seen in 3 patients (38%) and all had normal aortic root size.
Conclusion We describe long-term outcomes of patients with ALCAPA syndrome. Postoperatively, the majority remain asymptomatic with good exercise capacity. Their left ventricular systolic function is good. Life-long follow up is warranted.

10 LONG-TERM OUTCOMES OF ADULTS WITH WILLIAMS SYNDROME IN AN ADULT CONGENITAL HEART DISEASE CENTRE

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Background Williams syndrome, is a congenital, multisystem disorder, involving the cardiovascular, connective tissue and central nervous systems. We sought to determine long-term outcomes of these patients in our Adult Congenital Heart Disease Service.
Methods A retrospective review of case notes of patients with Williams syndrome was performed, reviewing clinic letters, operation details and cardiac imaging.
Results We identified 37 adults with Williams syndrome (62% male) with mean age 30±9.6 years (range, 19 to 56 years). 6 patients (16%) were discharged after their first review as no cardiovascular abnormalities were identified. One patient was lost to follow-up. Mean follow-up of the rest was 7.9±4.6 years (range, 6 months to 15 years). Most common cardiovascular manifestations included systemic arterial hypertension (40%, n=12), supravalvar aortic stenosis (57%, n=17; repaired (n=10)), pulmonary artery stenosis (30%, n=9; operated (n=5)) and aortic coarctation (20%, n=6; repaired (n=4)). At last clinic review, 10 patients (33%) were NYHA II (n=9) or NYHA III (n=1). 12 patients (40%) were hypertensive (BP >140 systolic), despite being on antihypertensive treatment (n=8). All patients were in sinus rhythm and no arrhythmias were identified. 9 patients (30%) had prolonged QTc (>440 ms in men or >460 ms in women). The majority (97%, n=29) had good left ventricular function (mean EF 63%±4.6%), and only one patient had mild impairment (EF 50%). No significant post-operative gradients were measured.
Conclusion Long-term follow-up of patients with Williams syndrome and significant cardiovascular disease is essential with particular care to blood pressure control.

11 PERSONALISED WARFARIN DOSING IN CHILDREN AFTER CONGENITAL HEART SURGERY: A RANDOMISED, PROSPECTIVE, CROSS-OVER, PILOT STUDY AT GLENFIELD HOSPITAL, LEICESTER

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After cardiac surgery (eg, Fontan repair, valve replacement), children are at an increased risk of thrombosis and therefore long-term oral anti-coagulation is essential to prevent morbidity and mortality. Warfarin is commonly used, but optimising the dose and maintaining a therapeutic INR is challenging for clinicians due to considerable inter- and intra-individual variability in its pharmacokinetics (PK) and pharmacodynamics (PD). The PK/PD is affected by both patient related (such as genetic polymorphisms of the metabolic enzymes) and environmental (eg, diet) factors. To improve the accuracy and consistency of warfarin dosing, a novel PK/PD (pharmacological) based model incorporating pharmacogenomics has been developed to assist clinicians in predicting initial and maintenance warfarin doses in post-operative cardiac children.[1]

The aim of the study is to compare warfarin dose management using pharmacological model with the traditional, 'trial and error' approach. The study is prospective and observational and involves 2 groups: In Group 1 (warfarin naïve) patients, loading and maintenance warfarin doses are estimated using the pharmacological model over 6 month duration and compared to historical case matched controls dosed according to the traditional approach. Group 2 patients already established on maintenance warfarin therapy entered a randomised cross-over study comparing pharmacological model-estimated dose adjustments with the traditional approach, over a 12 month period. The study also seeks to explore the views of children, parents and medical staff about the new model based approach.

The study commenced in October 2015 and recruitment stopped in December 2016. Group 1 (n=5) and Group 2 (n=29) participants are currently being followed up for their warfarin dosing and monitoring.

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12 IS ECHOCARDIOGRAPHY ALONE SUFFICIENT FOR RELIABLE CASCADE SCREENING OF FIRST DEGREE RELATIVES OF PATIENTS WITH BICUSPID AORTIC VALVES?

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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