Conclusion In the current analysis ESC/EAS guidelines are not the most frequently used tool for risk classification. Mean LDL-C levels both in high and very high risk patients are higher than the recommended goals and the prevalence of combination LLT therapy was low. Intensification of LLT in high and very high risk patients seem to be an unmet need in routine clinical practice in Ireland.

Introduction Hypertrophic cardiomyopathy (HCM) is the most common cause of sudden cardiac death in people < 35 years. To date, the estimation of risk has been based on clinical parameters such as LV outflow tract obstruction, ventricular arrhythmias, syncope and family history using risk prediction tools such as the ESC HCM SCD score. The use of genetics in prognostication or risk estimation is not well established. Using high-throughput sequencing technologies, higher proportions of complex genotypes, such as compound heterozygous, digenic patients and rare homozygous patients have been reported in up to 7% of cases. The impact of this complex genetic architecture on clinical care remains poorly defined. We present a case to highlight this complexity.

Background A 23 year-old white female was referred with a murmur. She had no past medical history and no family history of cardiomyopathy or sudden death. Her ECG showed sinus rhythm with subtle non-specific repolarization abnormalities. Echocardiography showed asymmetric septal hypertrophy with a maximum wall thickness of 23 mm in the mid-septum. An LV outflow tract gradient of 83mmHg post-Valsalva was recorded. Cardiac MRI demonstrated patchy late gadolinium enhancement in the hypertrophied inferoseptum. A stress test confirmed normal exercise capacity and haemodynamic response. A 24 hour Holter monitor showed one short run of atrial tachycardia and no ventricular arrhythmias or ectopics.

Management Risk stratification was undertaken using the ESC HCM Risk-SCD prediction tool. The score was estimated at 4.4% risk of suffering a life threatening event within 5 years. This value is in the intermediate range (4–5.9%) where an ICD can be considered but is not mandated. After a detailed discussion, the patient declined ICD implantation for cosmetic reasons. Genetic testing was performed. Two variants were identified in the MYBPC3 gene. The first was MYBPC3 c.3226_3227insT (p.Asp1076Valfs*6), a truncating Class 4 pathogenic variant. A second MYBPC3 missense mutation MYBPC3 c.1408C>T, p.(Arg470Trp) was also identified and
Early detection of late anthracycline cardiotoxicity in adult survivors of childhood malignancy

Background Almost 60% of the 2,000 children diagnosed with cancer in the UK and Ireland every year are treated with anthracycline chemotherapy. Anthracyclines are WHO essential medicines, and 5-year childhood cancer survival rates are approaching 85%, but also carry a risk of cardiotoxicity, particularly for childhood cancer survivors (CCS) of whom 30% have left ventricular dysfunction at 10 years and 10% have heart failure at 40 years post treatment. Current guidelines advocate periodic screening every 1–5 years however this approach is both resource intensive and inadequate, diagnosing cardiotoxicity at a late and often irreversible stage. Consequently, ‘Defining Robust Predictors of Cardiotoxicity’ was deemed a top ten priority for the cardio-oncology field at the Global Cardio-Oncology Summit.

Purpose Our research aims to investigate novel imaging and blood biomarkers (established, novel and biomarker discovery) to stratify childhood cancer survivors. Method Consenting adult patients will have cardiovascular risk factor screening, a quality-of-life questionnaire, six-minute walk test (6MWT) and a 24-hour Holter monitor. Patients will undergo 3D echocardiography, Global Longitudinal Strain (GLS) and Right Ventricular (RV) Strain imaging. Magnetic Resonance Imaging (MRI) will be conducted with feature tracking, myocardial tagging and aortic distensibility. Blood will be analysed for established (NTproBNP, CRP, and Troponin) and novel (IL6, sST2, MPO) biomarkers. Biomarker discovery through proteomic and microRNA analysis will be explored in subgroups of patients with normal ventricular function, impaired strain and ejection fraction. The top differentially expressed biomarkers will subsequently be validated within the remaining cohort.

Results 50 patients have been recruited to date (33 female, mean age 26). Half were treated before the age of 5. Mean dose was 265mg/m2 (120–480). Time since treatment was mean 18 years (median 16.5, range 6–33). 7 patients had a blood pressure >140/90mmHg, 18 had an abnormal lipid profile, and 1 had an abnormal HbA1c. Mean BMI was 27.1kg/m2 (18–49) with 23 participants classed as overweight/obese. 21 (42%) patients had a 6MWT distance below the lower limit of normal for their age/sex/height/weight. Left ventricular dysfunction was detected in 22/50 (44%): 3D ejection fraction was <55% in 7/46, GLS was abnormal in 22/43 and RV strain was abnormal in 15/31 patients. Troponin was normal in all patients. NTproBNP was elevated (>125ng/L) in 10 patients (20%). Elevated NTproBNP was associated with higher anthracycline dose (P=0.004, r=0.42). NTproBNP, CRP and Troponin were not associated with abnormal echocardiographic parameters. MRI and novel biomarker analysis will be conducted on enrolment completion.

Conclusion Childhood cancer survivors are at increased cardiotoxicity risk. Current results indicate that CCS have increased cardiovascular risk and impaired 6MWT. Established biomarkers fail to correlate with imaging data and as such support ongoing biomarker research.