AEPC-YIA Session

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[AEPCYIA] AEPC-YIA Session（II-AEPCYIA）

Chair:Jan Janoušek（Children's Heart Centre, Motol and Homolka University Hospital, Prague, Czech Republic）
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[II-AEPCYIA-4] Yield of Clinical Screening in Paediatric First-Degree Relatives of Patients With Desmosomal Cardiomyopathy

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Background and Aim:

Background and Aim: Variants in desmosomal genes (PKP2, DSP, DSG2, DSC2 and JUP), usually inherited as autosomal dominant traits, cause a range of arrhythmogenic cardiomyopathy (ACM) phenotypes, including arrhythmogenic right ventricular cardiomyopathy (ARVC), dilated cardiomyopathy (DCM), and non-dilated left ventricular cardiomyopathy (NDLVC). Historically, familial desmosomal cardiomyopathies have been considered adult-onset diseases, but the yield of clinical screening in paediatric first-degree relatives has not been systematically evaluated.

Methods:

Methods: Clinical data from all children (£18 years) referred to a single referral centre for family screening following a diagnosis of desmosomal cardiomyopathy in a first-degree relative were analysed.

Results:

Results: Of 173 children from 77 families referred for screening, 79 (47%) from 55 (71%) families harboured the familial desmosomal gene variant (26 DSP, 26 PKP2, 2 DSG2, 1 JUP). 11 children [14%; 6 females (55%)] from 8 families (10%) reached diagnostic criteria for one of the ACM phenotypes at a mean age of 12.5± 4.2 years, with 4 (5%) [JK2] younger than 10 years. 1 DSP and 1 PKP2 carrier each fulfilled diagnostic criteria for NDLVC (non-ischemic left vemtricular scar), 3 DSP carriers had DCM (of whom 2 were biallelic), and 6 PKP2 carriers had ARVC [Figure 1]. In addition, 2 patients had non-diagnostic features: 1 DSP carrier right ventricular (RV) hypokinesia on cardiac magnetic resonance (CMR,); 1 DSG2 carrier had borderline RV systolic impartment on CMR. Of the 6 PKP2-ARVC, 2 had non-sustained ventricular tachycardia (NSVT); 2 had RV dilation, 1 with aneurysms and RWMAs; 1 had ventricular ectopy (VE) >500/day. All DSP-DCM patients had extensive late gadolinium enhancement (LGE) and NSVT; both biallelic DSP-DCM patients underwent cardiac transplantation.

Conclusions:

Conclusion: Clinical screening identifies diagnostic criteria for an ACM phenotype (ARVC, DCM, NDLVC) in 14% of paediatric first-degree relatives of individuals with desmosomal-related

cardiomyopathy. These findings highlight the importance of commencing clinical screening at a young age.