

showed myocardial hypertrophy and granular appearance of the myocardium should be considered in the diagnosis of CA. Cardiac magnetic resonance imaging is valuable in the diagnosis of CA.

GW26-e2244

Gene Mutations in Chinese with Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy—a cohort registry study

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OBJECTIVES Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) is an inherited cardiac disease associated with an increased risk of arrhythmic sudden death. Mutations in desmosomal genes and some extra-desmosomal genes have been identified to associate with ARVD/C. Previously we identified 5 novel plakophilin (PKP2) mutations in a cohort of Chinese patients with ARVD/C. Our present study is to determine the prevalence of other associated gene mutations in this ARVD/C registry study and explore the potential genotype-phenotype relationship.

METHODS Genotypic and phenotypic profiles were studied in a cohort of 32 symptomatic Han Chinese with a clinical or suspected diagnosis of ARVD/C according to modified international Task Force criteria in 2010. Direct sequencing of 5 desmosomal genes and 3 extra-desmosomal genes was performed with a 3730XL DNA Analyzer.

RESULTS 22 mutations including 13 novel (13/22, 59.1%) in 5 desmosomal genes PKP2, Desmoplakin (DSP), Desmoglein-2 (DSG2), Desmocollin-2 (DSC2), Plakoglobin (JUP) were identified in 20 (20 of 32, 55.6%) patients in our cohort. No mutations were found in extra-desmosomal genes. Among 32 patients, 11 (11 of 32, 34.4%) patients have PKP2 mutations, 3 (9.4%) DSP, 3 (9.4%) DSG2, 6 (18.8%) DSC2 and 4 (12.5%) JUP. Multiple mutations were found in 6 subjects (6 of 32, 18.8%), among which 4 have PKP2 mutation, 3 DSC2, 3 DSP and 3 JUP. Genotype-phenotype analysis indicates compound multiple mutations may predict major structural abnormalities.

CONCLUSIONS PKP2 mutation is the most common gene mutations in our ARVD/C cohort. A higher percentage of DSC2 and JUP mutations were identified in the cohort compared with previous reports. Compound multiple mutations are common and may indicate major structural abnormalities. Extra-desmosomal genes mutations are rare in our Chinese ARVD/C cohort.

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Incremental Value of Contrast Echocardiography in the Diagnosis of Left Ventricular Noncompaction

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OBJECTIVES Contrast echocardiography with left ventricular opacification (LVO) can improve endocardium definition and potentially becomes supplement of conventional two-dimensional echo (2DE) in the diagnosis of noncompaction of the ventricular myocardium (NCVM). This study aimed to access the feasibility, accuracy, reproducibility of LVO & its incremental value than 2DE in NCVM diagnosis.

METHODS LVO & 2DE were performed in 85 patients (54 men, mean age 40±20 years) with suspected NCVM (NCVM Gp), and 2DE were performed in 40 healthy volunteers (Normal Gp, 20 men, mean age 40 ± 23 years). The LV chamber size and LV ejection fraction derived from Biplane Simpson's formula were compared among LVO-NCVM Gp, 2DE-NCVM Group & 2DE-Normal Gp. The location and extent of NCVM were evaluated based on AHA/ACA 16 segment model for LV segmentation, and the thickness ratio of noncompacted to compacted myocardium (NCR) were assessed on LVO & 2DE by 2 independently blinded experienced echo-cardiologists.

RESULTS (1) Compared with the Normal Gp, the NCVM Gp showed larger LVEDD (58.9±11.6mm vs. 45±5.1mm), LVEDV (121±11.5ml vs. 95±14.8), LVESD (43.0±3.44mm vs. 33±4.0mm), LVESV (74.3±3.94ml vs. 44±4.4ml), lower LVEF (40.8±13.2% vs. 65.6±7.1%) and E/A ratio (0.82±0.32 vs. 1.62±0.5) using 2DE method (p<0.05).

(2) Within the NCVM Gp, compared with the values from 2DE method, LVEDD (65.2±7.8mm vs. 58.9±11.6mm), LVEDV (162±14.8ml

vs. 121±11.5ml), LVESD (47.8±5.67mm vs. 43.0±3.44mm), LVESV (84.7±2.46ml vs. 74.3±3.94ml) derived from LVO method were larger and LVEF (38.2±12.4 vs. 40.8±13.2) on LVO was slightly lower (p<0.05).

(3) Among the whole 1360 LV segments in NCVM Gp, there were more segments adequately visualized for analysis on LVO than on 2DE (1278 vs. 1183, 93.97% vs. 86.99%). There were more noncompaction segments detected on LVO than on 2DE (314 vs. 262, 23.09% vs. 19.26%). Of the 921 segments interpreted as normal on 2DE, 52 segments (5.65%) were noncompacted on LVO. NCVM on LVO were majorly located in medium (53.18%), apical (46.18%) segments and lateral wall (39.81%); rarely involved in basal segment (0.64%).

(4) NCR on LVO was greater than that on 2DE (4.2±1.3 vs. 3.3±1.2, P<0.0001), but they are highly related and both showed excellent interobserver consistency. The coefficient of inter-observer variability of NCR was slightly smaller using LVO than 2DE (5.2% vs. 6.6%).

(5) The NCVM detection rate using 2DE, LVO, 2DE+LVO were statistically different, with the highest detection rate on 2DE+LVO and lowest detection rate on 2DE (89.4% vs. 63.5%, p<0.05).

CONCLUSIONS Contrast echo can clinically improve the diagnosis of NCVM in accuracy, sensitivity & reproducibility, and act as a useful supplement to the routine two-dimensional transthoracic echo.

GW26-e2395

A frame shift mutation(1208fs) in the MYBPC3 gene associated with hypertrophic cardiomyopathy in a Chinese family

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OBJECTIVES Hypertrophic cardiomyopathy (HCM) is the most common genetic cardiovascular disorder. This genetic diversity modifier genes form the basis of its phenotypic heterogeneity.

METHODS Clinical, three-dimensional speckle tracking (3D-STI), electrocardiographic (ECG), cardiac magnetic resonance (CMR) and echocardiographic(UCG) examination in members of a three-generation Chinese family was followed by exon and boarding intron analysis of 96 genes in the proband using second-generation sequencing. The identified mutations were confirmed by bi-directional Sanger sequencing in all family members and 300 healthy controls.

RESULTS The deleted mutation 3624 del C in exon 31 of the MYBPC3 gene were identified in proband and two family members(subjects I-2, III-1). While the remaining family and 300 normal controls did not find this mutation. The onset age proband is 42 years old and subject I-2 is 50 years old. Both of them were accompanied by chest pain. Subject III-1 UCG and CMR were normal. However his ECG showed sinus bradycardia and paroxysmal supraventricular arrhythmias. CMR results showed that proband has myocardial fibrosis in base-septal and anterior wall of left ventricular. Subjects I-2 has myocardial fibrosis in middle-septal, anterior wall and inferior wall of left ventricular. The mutation carriers showed septal, anterior wall, inferior wall and apical of left ventricular hypertrophy in UCG compared with the no-mutation carriers, in the family; In 3D-STI, the global and the segmental longitudinal strain in middle of the posterior interventricular septum, the base and the middle of anterior interventricular septum, and the middle of the left ventricular anterior wall were reduced. Furthermore the segmental area strain in the base of the anterior interventricular septum were reduced (P <0.05).

CONCLUSIONS We demonstrate a close correlation between clinical phenotype and genotype of MYBPC3 gene Pro1208fs mutation in a Chinese family with HCM for the first time. The mutation results in 100% penetrance. Mutation carriers are late age of onset and no specific clinical symptoms. Partial myocardial fibrosis and Maron III type hypertrophy and a high potential of abnormal myocardial systolic function were detected suggesting the pathogenesis of the mutation.

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A TNNT2 gene mutation associated with dilated cardiomyopathy in the Xinjiang Kazak ethnicity

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OBJECTIVES Dilated cardiomyopathy (DCM) is characterized by left ventricular enlargement, systolic dysfunction, and heart failure. Both genetic and non-genetic factors have been linked to DCM pathogenesis. Familial DCM (FDCM) accounts for 20%-50% of all DCM cases, highlighting the importance of genetic in pathogenesis. Indeed, more than 40 DCM