GW26-e3571
Echocardiographic Diagnosis of Rare Pathological Patterns of Sinus of Valsalva Aneurysm

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OBJECTIVES Echocardiography is the first choice for the diagnosis of sinus of Valsalva aneurysm (SVA). However, operator inexperience to rare pathological patterns makes it easy to misdiagnose or fail to diagnose SVAs.

METHODS Echocardiographic features and surgical findings of 270 Chinese SVA patients treated in the last 18 years (1995-2013) at the Union Hospital were compared retrospectively, of which 22 cases were rare patterns.

RESULTS The patients with a rare origin, a rare extending position and a rare course accounted for 3.4%, 7.4% and 0.4% of 270 cases, respectively. The three most common aneurysmal complications of these patients with rare patterns were severe aortic regurgitation (16), obstruction of the ventricular outflow tract or valvular orifice (3) and conduction disturbance (3). The origin, course, extending position and rupture status of the SVA determined by echocardiography were entirely consistent with surgical findings in 81.8% of 22 cases. With the exception of one failed diagnosis of an aneurysmal wall dissection and one misdiagnosis of a descending aortic dissection, the echocardiographic results of SVA complications and associated cardiovascular lesions were also confirmed.

CONCLUSIONS Echocardiography can accurately diagnose SVAs with different rare pathological patterns by identifying distinguishing features. However, for several conditions, echocardiography alone could not accurately identify the origin or course of the aneurysm or define its relationship to adjacent structures. Therefore, combining different imaging techniques, such as CT angiography and aortic angiography, is recommended.

GW26-e1532
Targeted Next-Generation Sequencing on Sporadic Thoracic Aortic Aneurysm and Dissection Individuals: Discover the Genetic Connections

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OBJECTIVES Thoracic aortic aneurysm and dissection (TAAD) has become a clinical emergency situation that threats human life seriously during past decades in China. Hypertension and hyperlipoidemia are considered as the high risks for TAAD. Genetic studies have revealed that mutations on certain genes might be the real cause for inherited TAAD, such as mutations in FBN1, TGFBR1/2, and SMAD3. Next-generation sequencing is now widely used to find possible genetic changes for familial TAAD patients and to help them reducing mortality events. However, most next-generation sequencing data requires professional bioinformatics analysis. Moreover, most patients have been proved without any known familial risks for TAAD. Limited genetic associations are found among sporadic TAAD individuals. This study sought to apply a targeted next-generation sequencing panel with associated genetic information to 2 TAAD families and 8 sporadic TAAD individuals to unravel the potential genetic connections among sporadic TAAD.

METHODS We applied a targeted next-generation sequencing panel for 4813 genes including most reported genes associated with TAAD. Screens were performed in 2 families, both of which were with one member having TAAD, and 8 unrelated individuals with TAAD. Sequencing data was analyzed based on the information provided in the panel. Extensive cardiological examination was performed, which included physical examination, electrocardiography, and echocardiography.

RESULTS Two potential novel variants on FBN1 and MMP12 respectively were indentified in one family. Those two variants were only found in the proband, not in any other family members. Among the other family and 8 sporadic TAAD individuals, neither 2 potential novel variants nor any other reported mutations were detected.

CONCLUSIONS Our finding suggests that next-generation sequencing panel targeted on 4813 genes is a promising tool for early genetic diagnosis of TAAD from known genes. With all the information in the panel, clinicians may provide genetic consultations within days. Despite the advantages above, few associations between TAAD individuals in our study implies that entire exon sequencing on larger population may provide more information for targeted sequencing panel and reveal the underlying genetic pathogenesis for TAAD.

GW26-e1832
CYP4F2 genetic polymorphisms are associated with coronary heart disease in a Chinese population

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OBJECTIVES To explore the relationship between CYP4F2 gene polymorphism and coronary heart disease (CHD) in a Chinese Han population.

METHODS We selected 440 CHD patients and 440 control subjects to perform a case - control study. Four SNPs (rs2108622, rs3093100, rs3093105 and rs3093135) in CYP4F2 gene were genotyped using polymerase chain reaction - restriction fragment length polymorphism (PCR - RFLP) methods. The genotype and haplotype distributions were compared between the case and the control group.

RESULTS We found both rs2108622 and rs3093105 in CYP4F2 gene were associated with the risk for CHD (P $<$0.01). Haplotype analysis indicated that GGGT haplotype consisted by rs2108622-rs3093100-rs3093105-rs3093135 was associated with CHD risk (OR = 4.367, 95% CI: 2.241 $\sim$ 8.510; P $<$ 0.001), but GGTA haplotype was associated with decreased risk for CHD (OR = 0.450, 95%CI: 0.111 $\sim$ 0.777; P $<$0.001).

CONCLUSIONS CYP4F2 gene polymorphisms were associated with the risk of CHD in Chinese population.

GW26-e4563
Serum Level of Fibroblast Growth Factor 21 is Independently Associated with Acute Myocardial Infarction

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OBJECTIVES Fibroblast growth factor 21 (FGF21) has been described a metabolic hormone critical for glucose and lipid metabolism. Previously, high levels of FGF21 were observed in patients with coronary heart disease and non-acute myocardial infarction (non-AMI). In this study, we investigated the changes in FGF21 levels in Chinese patients with AMI.

METHODS We enrolled 55 patients in whom AMI (including ST-segment and non-ST-segment elevation, within 24 h after admission) was diagnosed between March and August 2013 at the Ministry of Health, Beijing Hospital. The control group included 45 patients with chest pain without creatine kinase (CK), creatine kinase- MB (CK-MB) and Troponin T (TNT) elevation. The patients in control group were also diagnosed coronary heart disease by Coronary angiography. We used ELISA to measure circulating FGF21 levels in 55 AMI patients and 45 non-AMI control patients on the 1st day after syndrome onset. All patients were followed-up within 30 days.

RESULTS FGF21 levels in AMI patients were significantly higher than those in non-AMI controls (0.25 (0.16-0.34) vs. 0.14 (0.11-0.20) ng/mL, P $<$ 0.001). FGF21 levels reached the maximum within approximately 24 h after the onset of AMI and remained at high for 7 days, and the FGF21 level (OR: 16.93; 95% confidence interval (CI): 2.65-108.05; P $=$ 0.003) was identified as an independent factor associated with the presence of AMI. On the 7th day, FGF21 levels were significantly higher in the patients who subsequently developed re-infarction within 30 days than in the patients who did not develop re-infarction (with vs. without re-infarction: 0.45 (0.22-0.64) vs. 0.21 (0.15-0.29) ng/mL, P $=$ 0.014).

CONCLUSIONS The level of serum FGF21 is independently associated with the presence of AMI in Chinese patients. High FGF21 levels might be related to the incidence of re-infarction within 30 days after onset.