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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 acute aortic regurgitation (16), obstruction of the ventricular outflow track 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 threatens human life seriously during past decades in China. Hypertension and hyperlipidemia 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 on FBN1, TGFB1, 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-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. Previous studies showed that high levels of FGF21 were associated with the risk of CHD (P < 0.01). Haplotype analysis indicated that GGGT haplotype consisted by 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 ~ 8.510; P < 0.001), but GGTA haplotype was associated with decreased risk for CHD (OR = 0.450, 95%CI: 0.111 ~ 0.777; P < 0.001).

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