Case report of ventricular septal hypertrophic cardiomyopathy and left ventricular non-compaction carrying OBSCN and FHOD3 gene mutations

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Introduction: A 19-year-old male was admitted to our hospital because of syncope. He denied any history of hypertension and diabetes. A little brother of his died of suspicious cardiac causes at 3 months old of age and other family members are healthy. He underwent multiple electrophysiology studies (EPS) since 4 months of age, during which he was diagnosed with pre-excitation syndrome with multiple atrio-ventricular accessory pathways and dual atrioventricular nodal pathways, which was all successfully treated by ablation. Meanwhile, echocardiogram showed “symmetrical hypertrophic cardiomyopathy and normal ejection fraction”. He stopped regular follow-ups 5 years ago because there were no more symptoms. Activity-related palpitations occurred again and then developed syncope during running. EKG showed wide QRS tachycardia (figure 1&2)

Methods: By using cardiac imaging tools, genetic sequencing methods and EP study, we define the diagnosis of this young patient

Result: EPS showed fasciculo-ventricular pathways which was inconsistent with the wide QRS tachycardia and dual atrioventricular nodal pathways which failed to induce tachycardia, therefore he didn’t undergo ablation. In hospital cTNI and CK-MB was elevated. Echocardiogram (figure 3,4&5) showed an enlarged left ventricle, severely reduced left ventricular ejection fraction, and unsymmetrical septal hypertrophy and left ventricular non-compaction (LVNC), while the so-called symmetrical left ventricular posterior wall hypertrophy turned out to be LVNC. Besides the echo manifestations, Cardiac magnetic resonance showed a decrease perfusion of left ventricular middle layer myocardium, and late gadolinium enhancement of left ventricular middle layer and subepicardial myocardium. Genetic testing showed he was a carrier of variants p.Val5836Met of the OBSCN gene and variants p.Met1212Thr of the Formin Homology 2 Domain Containing 3 (FHOD3) gene. Family genetic sequencing showed his father and old sister were both carriers of these two variants while his mother wasn’t, but all their echos and EKGs were normal. In summary, ventricular septal HCM and LVNC was diagnosed. We prescribed him sacubitril valsartan, bisoprolol and spironolactone to treat heart failure and warfarin to prevent thromboembolic events. Since there were no pacing indications, subcutaneously ICD was implanted to prevent SCD (figure 6). Now he is on follow-ups and has no syncope episodes, thromboembolic events and heart failure symptoms

Conclusion: LVNC is easily misdiagnosed as HCM and we should carefully use cardiac imaging tools to differentiate. It can be combined with other cardiomyopathies such as HCM/DCM. In our case,
LVNC and septal HCM is presented in one patient and his symptoms were mainly arrhythmia-related. And we identified a novel gene mutation of the FHOD3 gene in his family which needs more evidence in future studies.