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LVNC – A review

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Abstract

LVNC is a relatively new clinical entity, with a significant increase in awareness and diagnosis in recent years. Currently the aetiology and pathogenesis of LVNC remains uncertain, alongside prevalence, however the diagnosis of LVNC appears to be increasing with improving imaging techniques.

For educational purposes involving a rare clinical condition, we present the case of a 52 year old gentleman who was diagnosed with LV non compaction via ECHO and CMR. Interestingly it was noted two of his children had congenital heart disease, one daughter had Tetralogy of Fallot, and a second daughter had both an ASD and VSD.

Challenges facing LVNC involve difficulty of diagnosis with no gold standard yet available, uncertainty of benefit with standard disease modifying therapies for HF-REF, and apparent increased risk of arrhythmias suggesting early ICD placement may be warranted for patients.

Keywords: Hr-Ref; heart failure; lv non compaction; arrhythmias; lcd Risk.

Abbreviations: LVNC: Left Ventricular Non Compaction; HF-REF: Heart Failure With Reduced Ejection Fraction; HF: Heart Failure; CCU: Coronary Care Unit; ACS: Acute Coronary Syndrome; DAPT: Dual Antiplatelet Therapy; VT: Ventricular Tachycardia; NSVT: Non Sustained Ventricular Tachycardia; HCM: Hypertrophic Cardiomyopathy; DCM: Dilated Cardiomyopathy; ICD: Implantable Cardiac Defibrillator; CMR: Cardiac MRI; LGE: Late Gadolinium Enhancement; TOF: Tetraology Of Fallot; TGA: Transposition Of The Great Arteries; HLHS: Hypoplastic Left Heart Syndrome; ASD: Atrial Septal Defect; VSD: Ventricular Septal Defect; PDA: Patent Ductus Arteriosus; LVEF: Left Ventricular Ejection Fraction; AF: Atrial Fibrillation; TIA: Transient Ischaemic Attack.

Introduction

We present the case of a 52 year old gentleman diagnosed with an unusual aetiology of HR-REF, being LV non compaction.

Case presentation

A 52 year old male presented to our emergency department with one day of chest pain, and two weeks of rapidly progressive shortness of breath.

On arrival he was hemodynamically stable and examination revealed elevated JVP with bilateral inspiratory crepitations. ECG showed sinus rhythm with LBBB, and CXR cardiomegaly and upper lobe diversion. Admission bloods showed a significantly elevated troponin at 1500 and NT-proBNP at 3800, remaining bloods were normal.

He was admitted to CCU with concern for ACS complicated by acute decompensated heart failure, and managed with DAPT, heparin and intravenous diuresis.

He had frequent runs of monomorphic VT up to 40 seconds on telemetry monitoring. Coronary angiography showed normal coronaries. Echocardiography showed a severely dilated LV, and severely depressed LVEF of 15% with trabeculation and ventricular recesses suspicious for LV Non Compaction (LVNC). CMR was arranged and reported findings consistent with LVNC.

He was commenced of heart failure modifying medications. NSVT and VT persisted on bisoprolol and therefore an ICD was inserted. He was commended on NOAC therapy for prevention of thromboembolism, and discharged home for HF follow up.

Interestingly it was noted two of his children had congenital heart disease, one daughter had Tetralogy of Fallot requiring corrective surgeries soon after birth. A second daughter had both an ASD and VSD closed in early childhood.

Discussion

LVNC is a relatively new clinical entity, with an increase in awareness and diagnosis in recent years. It was first described in association with congenital heart disease by Grant in 1926, noting appearances of a "Spongey Myocardium" in some patients. Descriptive cases progressed, and further reports in the 1970's and 1980's noted isolated cases of heart failure with persisting embryonic myocardium [1,2].

The term '*isolated non-compaction of left ventricular myocardium*' was introduced by Chin et al in 1990 [3], which recognised LVNC as a rare disorder of endomyocardial morphogenesis with prominent ventricular trabeculations and deep intertrabecular recesses.

Currently, the exact aetiology and pathogenesis of LVNC remains uncertain, but it suspected due to an arrest in endomyocardial morphogenesis with failure of compaction. Trabeculation is seen at 4-8 weeks of cardiac embryological development, likely to increase surface area without an effective epicardial coronary circulation. Compaction begins around 8 weeks gestation, coinciding with the growth of the epicardial circulation, progressing from epicardium to the endocardium, from the base to the apex, and the septum to the free wall, therefore an arrest in the process almost always involves the apex [4].

LVNC can occur in isolation, or as part of a syndrome with other congenital heart disease including TOF, TGA, HLHS, ebsteins anomoly, ASD and VSD, PDA, bicuspid aortic valve and coartation of the aorta [5]. It has also been described in association with metabolic diseases such as Barths Syndrome, and neuromuscular disorders.

The AHA recognise LVNC as a genetic cardiomyopathy, while the WHO and ESC label LVNC as an "unclassified cardiomyopathy". Genetic information so far shows LVNC can be inherited most commonly in an autosomal dominant manner [6], alongside autosomal recessive and X linked transmission. A genetic mutation can be identified in 17-41% of patients screened, depending on patient selection and degree of genes screened [7].

The most commonly identified mutations occurs in sarcomere genes MYH7, MYBPC2 and TTN (71% of genetically positive patients) [8]. Other mutations identified affect cytoskeletan and signalling proteins, and overlap is seen with other congenital diseases as the same gene defects can be seen in HCM and DCM. Children are more likely to have an identifiable genetic cause, and can have complex genotypes with 2 or more mutations usually resulting in earlier onset and more severe phenotype [8]. 52% of 327 patients assessed in this review had a sporadic mutation with no family history.

The prevalence of LVNC is uncertain but appears to be increasing. Estimates vary 0.014 to 1.3% [9]. It can be found incidentally without any phenotypical disease in up to 8% of athletes [10], in sickle cell anaemia, and in pregnancy where it can regress without complication post natal. It remains uncertain if all cases of LVNC are genetic or can be acquired and develop in later life, as some case reports describe patients with previously normal echocardiograms and later development of LVNC.

LVNC can present in childhood usually with severe heart failure. Adult presentations frequently involve the triad of heart failure, arrthythmia (AF or ventricular) and thromboembolism.

Diagnosis can be difficult and echocardiogram is the first choice modality. Findings suggestive of LVNC on echo include prominent trabeculation of the ventricle with deep intertrabecular recesses, and the appearance of two layers to the myocardium. Criteria have been developed to increase diagnostic accuracy, the main three in use being the Chin, Jenni and Stollberger criteria. None are considered gold standard, and up to 8% of control healthy populations can meet requirements for diagnosis particularly black people [11], suggesting they are too sensitive.

CMR is increasingly being used to aid diagnosis. Peterson et al and Jaqueir et al looked at the accuracy of CMR in identifying LVNC in patients, and found a ratio between non-compacted and compacted layer >2.3, and trabeculated LV mass >20% of the global LV mass, have high levels of sensitivity and specificity to detect LVNC. As CMR progresses alongside disease understanding, parameters may be updated and improved upon [12]. LGE did not accurately discriminate for LVNC, however the presence of LGE suggests a lower LVEF and increased risk of tachyarrhythmias [13].

HF should be managed as per standard HF guidelines. General consensus suggests anticoagulation as per other recommendations and if LVEF is <40% as there is a risk of thromboembolism from intracavity abscesses. In one study, 10.8% of patients had identifiable thrombus on ECHO, and 33% had TIA/stroke without AF detected [16]. Regular screening of patients for arrhythmia with 48 hour Holter monitoring should be considered as VT has been detected in up to 35% of patients [16], and ICD placement is recommended with LVEF <35% and NYHA class II-IV. Suggestions have been made to consider a risk calculator similar to the HCM risk assessment, as these patients have a tachyarrthymia risk out of proportion to LVEF.

Patient prognosis depends upon disease phenotype. Initial reports suggested a very high mortality up to 35% over 3.7 years [14], but further information and case studies suggest this is an overestimate and now mortality reports range 2-20% [15]. A study in the EHJ note that the risk of CV death, VT, appropriate ICD shock or CPR was higher with LVNC then in an age matched cohort of DCM patients and noted the LVEF was higher in the LVNC group, therefore risks do not solely correlate to LVEF [16]. Adverse prognostic markers include a higher NYHA class, LV systolic dysfunction, and LGE on CMR. Cardiac transplantation should be considered for refractory heart failure or arrhythmia.

Conclusion

LNVC is a relatively rare condition with increasing recognition in recent years. Individual case reports and case series suggest patient outcomes are poorer and risks of sudden cardiac death are higher than in DCM with similar ejection fractions, and as such further information in the area is required, alongside consideration of developing a risk calculator similar to HCM for ICD placement.

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