Mrs K., a 55 years old female presented with gradual onset breathlessness of 5 years duration and had now paroxysmal nocturnal dyspnoea. Recently she had been getting palpitations and would feel orthopnoic at times. She also had transient right sided weakness with dysarthria which improved spontaneously within few days and for this she did not seek any medical advice. She had developed mild pedal edema. Physical examination revealed pulse 90/min, BP 110/70, respiratory rate of 23/min. JVP 7 cm above sternal angle. Heart was 2 cm below right costal margin with tenderness on deep palpation. There was pedal edema. Chest auscultation revealed bibasal rales. On cardiac examination there was cardiomegaly. Soft S1 and S2 with prominent S3. Grade 3 systolic murmur with preferential radiation to axilla. She had no focal neurological deficit at present.

In ECG there was sinus tachycardia, signs of LA enlargement, left bundle branch block (LBBB). Chest X-Ray showed cardiomegaly with prominent hila and blunting of right costophrenic angle. Echocardiogram revealed dilated left atrium and coronary sinus. LV was also dilated with prominent trabeculations in apical segments and mid lateral segments with free to-and- fro flow in intertrabecular recesses on color Doppler. Ratio of trabeculated layer (noncompacted) to compact layer was >2.5 as calculated in short axis view. No thrombus was seen in any chamber. LV ejection fraction was 36% by Simpsons method. There was grade 3 mitral regurgitation. No gradient across aortic valve. Right ventricular systolic pressure estimated from tricuspid regurgitation jet was 36+right atrial pressure.

Echocardiography consistent with LV noncompaction (See Fig. 1)

Patient was started on Toresamide, Spironolactone, Enalapril. Carvedilol was also started in low doses which she tolerated well. Patient was also anticoagulated with Warfarin.

Left Ventricle Noncompaction.

Left Ventricle Non Compaction (LVNC) described recently is congenital cardiomyopathy characterized by distinctive spongy appearance of LV. There are prominent trabeculations in left ventricle with intertrabecular recesses/ sinusoids continuous with ventricular cavity and have no connection with coronary vessels. There may be isolated non-compaction of left ventricle (INLV) (1) or may be associated with other cardiac abnormalities (2, 3). European Society of cardiology has grouped it under unclassified cardiomyopathy (3). American Heart association has classified it as primary genetic cardiomyopathy (9). Prominent trabeculations with spongy appearance is normal in early development of heart in fetus. It persists partly in right ventricle but there is regression of trabeculations in left ventricle and by birth LV has a compact wall with smooth luminal surface. Trabeculation clearance starts from basal segments and are last to clear from apical and lateral wall. Failure of regression of these trabeculations leads to left ventricular noncompaction (LVNC) (4). Some cases of LVNC is may be familial with autosomal dominant inheritance. Some cases are related to mutation in genes including ZASP (6), dystrobrevin and taffazin (7). Barth syndrome (BTHS) was reported in 1983 presenting as cardiomyopathy, neutropenia and skeletal myopathy. Disease is related to tafazzin gene on X chromosome that leads to deranged phospholipids metabolism.(8).They present with congestive heart failure and have noncompaction of left ventricle (LV)(7).
First detailed report isolated left ventricle noncompaction (LVNC) was of that of Chin TK et al 1990 who gave follow up of 8 cases including 3 necropsies. They proposed and used echocardiographic criteria for diagnosis of noncompaction. They also noticed some facial abnormalities in 3 of their cases and familial occurrence in 4 cases (1).

Ritter and colleagues (10) reported a series of adults with isolated ventricular noncompaction (IVNC), including many asymptomatic patients who were identified through routine echocardiographic evaluations. A prevalence of isolated ventricular noncompaction was found to be 0.05% in their echocardiographic series. From 1984 to 1993 they had 37,555 echocardiographic procedures and detected 17 cases of isolated ventricular noncompaction (IVNC). Prognosis in the asymptomatic patients in their study was clearly better than the prognosis in the symptomatic patients, with a mean follow-up duration of 30 months.

In a nationwide study in Japanese children by Ichida F et al in 150 hospitals(11) children one week to age of 15 years were screened and they had 27 patients of LV noncompaction. They were followed for 17 years. As most of patients were at asymptomatic stage they had good prognosis with late and gradual depression of LV function. 6 patients had signs of heart failure at presentation. 22 (88%) had ECG changes in form of T inversions in inferior and/or chest leads. 4 (18%) patients had WPW type pre-excitation 1 of which had concealed pathway clearly more prevalent than in normal population. Familial occurrence was noted in 12 patients.

Jenni R. et al (21) studied myocardial blood flow in 12 patients of LVNC as compared with normal subjects. They found that in patients with LVNC, a decreased coronary flow reserve is not confined to noncompacted segments, but extends to most segments with wall motion abnormalities. Thus, coronary microcirculatory dysfunction is associated with LVNC. Pignatelli RH et al (32) retrospectively reviewed 36 children with LVNC in Texas Children’s Hospital from 1997 to 2003. Age was from 1 day to 17 years. Average follow up was 3.2 years. There were 16 females and 20 males. What they found is that LVNC does not have an invariably fatal course when diagnosed in the neonatal period. Many have transient recovery of function followed by later deterioration, which may account for many patients presenting as adults, some manifesting an "undulating" phenotype.

Largest reported follow up is provided by French registry published in European J of Heart Failure (12). Out of 154 patients of suspected LVNC 105 were confirmed with definite diagnosis of isolated left ventricular noncompaction (LVNC) in central core laboratory screening. They comprised 69 (66%) men and 36 (34%) women aged 18–86 years (mean 45±17 years). Twenty-five (24%) patients were aged 60 years or more at the time of echocardiography and 14 were aged 70 years or more. 18 cases had familial history. It was also observed that Left ventricular non-compaction was frequently over-diagnosed by echocardiography. Although familial screening revealed 8% cases were asymptomatic, patients identified as LVNC presented with a high risk of severe complications, transplantation or death and needed close follow-up.

Clinical features

Until recently, isolated LVNC was thought to be extremely rare with prevalence of 0.3% in adults (10, 17, 20) and an annual incidence of, 0.1 per 100 000 in children. (18,19, 20). But as of now incidence seems to be higher possibly due to better awareness, relatively sensitive criteria used and better diagnostic equipment. In a recent study (20) unexpectedly high percentage of patients with heart failure were noted fulfilling current echocardiographic criteria for LVNC. This might be explained by a hitherto underestimated cause of heart failure, but the comparison with controls suggests that current diagnostic criteria are too sensitive. Many cases or picked up accidentally on echocardiographic examination. Symptoms may develop at any stage from early childhood. About one fourth of cases are more than 60 years of age and cases are seen in up to 8th decade of life (12). Disease is seen in both males and females. Prognosis depends on symptomatic state of patient. More the patient is symptomatic worse the prognosis. Most of patients present with signs of congestive cardiac failure (43%). In a large European study a significant number of cardiac failure patients when screened fulfill criteria for LV noncompaction (20). Some present with features of dilated cardiomyopathy. Other manifestations are arrhythmia (11%) and embolism (4%). Angina has also been reported in LV noncompaction (15). 17% cases have familial history (12). Electrocardiogram commonly shows non specific T wave changes in inferior as well as lateral leads and usually has left bundle branch block (LBBB) pattern in adults (12). Increased incidence of WPW type pre excitation was noted in younger patients in a Japanese study (11).
Diagnosis

Diagnosis of isolated noncompaction of left ventricle is based on echocardiographic findings though MRI(22), and CT have also been used (11). Different echocardiographic criteria have been used for diagnosis of noncompaction.

1. Chin TK et al (1)

LVNC is defined by a ratio of X/Y ≤ 0.5

X = distance from the epicardial surface to the trough of the trabecular recess

Y = distance from the epicardial surface to peak of trabeculation

These criteria focus on trabeculae at the LV apex on the parasternal short axis and apical views, and on left-ventricular free-wall thickness at end-diastole

2. Jenni et al. (13)

(i) A two-layer structure, with a thin compacted layer and a thick non-compacted layer measured in end systole at the parasternal short-axis views

LVNC is defined by a ratio of N/C ≥2 where

N = non-compacted layer of myocardium

C = compacted layer of myocardium

(ii) Absence of co-existing cardiac structural abnormalities

(iii) Numerous, excessively prominent trabeculations and deep intratrabecular recesses

(iv) Recesses supplied by intraventricular blood on colour Doppler

3. Stollberger et al. (14)

(i) More than three trabeculations protruding from the left-ventricular wall, apically to the papillary muscles, visible in a single image plane in diastole

(ii) Intertabecular spaces perfused from the ventricular cavity, visualized on colour Doppler imaging:

Trabeculations are seen mainly in apical segments also in mid lateral and mid inferior segments of LV. Less commonly in mid anterior and mid septal segments. It is rarely seen in basal segments. There are contradictory reports regarding correlation of severity of trabeculations and symptomatic state of patients. Largest report of French registry did not show any correlation. In the recent study by Belanger et al., (16) a classification scheme was proposed, based on the assessment of the magnitude of the Non Compaction/Compaction ratio and the extent of the left ventricular non-compaction. They found that the degree of extension of LV noncompaction was related to the severity of LV dysfunction.

Treatment

Treatment depends on manifestations. Asymptomatic patients need just follow up. Patients presenting with cardiac failure are treated with usual regimen of Ace-inhibitors/Angiotensin blockers, Beta blockers, Diuretics. Some patients may need anticoagulants and anti-arrhythmic medication. Cardiac synchronization therapy, ICD and even cardiac transplantation have also been used.

References:


Conflict of Interest: None.

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