Isolated Left Ventricular Noncompaction: an underestimated cardiomyopathy?

M Banci, R Martinoli, S Piccirilli, L Santucci, A Dofcaci, F Papetti, I Sansoni, P Saccucci

Citation

M Banci, R Martinoli, S Piccirilli, L Santucci, A Dofcaci, F Papetti, I Sansoni, P Saccucci. *Isolated Left Ventricular Noncompaction: an underestimated cardiomyopathy?*. The Internet Journal of Cardiology. 2008 Volume 7 Number 2.

Abstract

Sir

Isolated Left Ventricular Noncompaction (IVNC) is a rare congenital unclassified cardiomyopathy. This disorder, become widely known in the last 10 years, is postulated to be caused by intrauterine arrest of compaction of the myocardial fibres and meshwork, an important process in myocardial development. The predominant clinical features are heart failure, arrhythmias, embolic events and unexpected death (Jenni R et al., 2001) [1]. By definition, IVNC occurs in the absence of other structural heart disease. According to the new American Hearth Association classification of the cardiomyopathies, IVNC is classified as a primary, genetic cardiomiopathy but in the position statement of the European Society of Cardiology it is not yet considered as a distinct pathology (Elliott P et al., 2008) [2]. There is evidence that IVNC is a genetically heterogeneous disease. Using linkage and mutation analysis, a point mutation in the G4.5 gene has been identified in a family with X-linked infantile IVNC and in other forms of infantile X-linked dilated cardiomyopathies. Sabine Sasse-Klaassen et al., (2004) suggest an autosomal dominant mode of transmission of this disorder in adult subjects that seems to be distinct from paediatric form. A genome-wide linkage analysis of a family with autosomal dominant IVNC showed a gene locus that maps to human chromosome 11p15 (Sabine Sasse-Klaassen et al., 2004) [3]. Nevertheless, a major genetic cause for familial IVNC remains to be identified. The true prevalence of IVNC as a cause of heart failure and heart transplantation is unknown. In a recent study of primary cardiomyopathy in Australian children, IVNC was the third most common cardiomyopathy after dilated cardiomyopathy and hypertrophic cardiomyopathy and in another single center, IVNC was responsible for 9,5% of cardiomyopathies in children (Pignatelli RH et al., 2003) [4]. Recently, Kovacevic-Preradovic's group in a retrospective study of 960 patients seen in the heart failure clinic from 1987 to 2005, describe the IVNC as a cause of heart failure common to hypertrophic cardiomyopathy, present in 2,7% of patients.

In our Center, Valmontone Hospital, in one year and half we have collected 132 patients with IVNC on 5570 heart failure, diagnosed with doppler echocardiography according to the criteria proposed by Jenni et al., 2001 [1]: 76 males and 56 female from the same Caucasian population with medium age of 40, range 5 months-86 years (Table 1).

Figure 1

Tab. 1. Aetiology of heart failure in our Center (Valmontone Hospital).

	All patients (n=5570)	Men (n=3222)	Women (n=2348)
CAD	2426 (12,89)	1500 (46,55)	926 (39,44)
Dilated cardiomyopathy	568 (10,20)	360 (11,17)	208 (8,86)
Valvular heart disease	514 (9,23)	224 (6,95)	290 (12,35)
Congenital heart disease	140 (2,51)	66 (2,05)	74 (3,15)
IVNC	132 (2,37)	76 (1,74)	56 (3,24)
Hypertensive heart disease	1300 (23,34)	700 (21,72)	600 (25,55)
Hypertrofic cardiomyopaty	450 (8,08)	300 (9,31)	150 (6,39)
Myocarditis	10 (0,18)	4 (0,12)	6 (0,25)
Amiloid Heart disease	8 (0,14)	2 (0,06)	6 (0,25)
Idiopathic restrictive cardiomyopathy	6 (0,10)	0 (0,00)	6 (0,25)
Arrhythmogenic right ventricular cardiomyopathy	8 (0,14)	4 (0,12)	4 (0,17)
Neuromuscolar disease	8 (0,14)	6 (0,19)	2 (0,08)

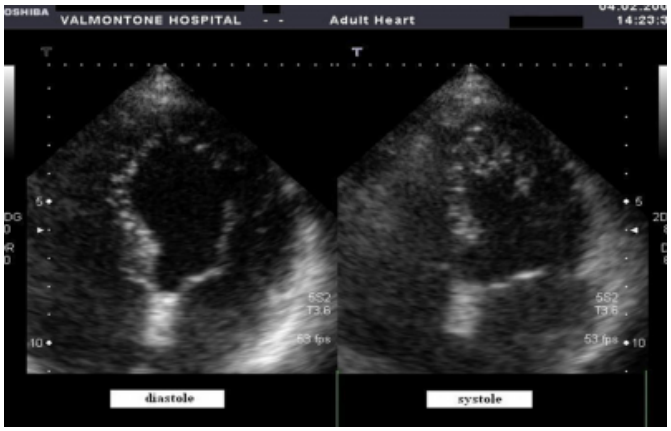
Patients or their parents, in case of <18 years, gave consent for participation in the study that was approved by the IRB. The diagnosis of IVNC was confirmed by three different

operators.

The Figure 1 shows an apical four chamber trasthoracic echocardiogram showing prominent trabeculation and intertrabecular spaces affecting the apex and lateral walls of the left ventricle.

Figure 2

Fig. 1. Apical four chamber trasthoracic echocardiogram showing prominent trabeculation and intertrabecular spaces affecting the apex and lateral walls of the left ventricle.



In our patients, we observed that the paediatric form of the disease has a different course and it is phenotipically distinct from the adult form, even if the anatomic alterations are apparently the same. Surely, the partitioned distribution is decisive: if the number of the involved segments is elevated the prognosis will be worst. In our cases we have observed furthermore an interesting correlation between face alterations and the disease's severity (data not shown).

The paediatric form is more severe than the adult form, while the adults have often lowest symptoms because of other therapies in progress which give them a protection from possible malformations. Therefore our opinion is that it is important to identify as soon as possible the start of symptoms and/or systolic/diastolic dysfunctions to begin the correct therapy and to prevent the disease progression. A higher case study and a long term follow-up will lead to implement non-morphological criteria of IVNC and to understand the treatment and the course of a disease still now not well clarified. Moreover, early and accurate diagnosis of INVC is important because the clinical manifestation is characterised by severe morbidity and mortality caused by heart failure, life threatening ventricular arrhythmias, and systemic embolic events (Ritter M et al., 1997) [5].

In reason of the relatively short time taken to collect our cases, our opinion is that this pathology is actually underestimated. The unavailability of high-performance echocardiography machines, the lack of operators trained in the IVNC's diagnosis, and the difficulty to suspect this pathology in patients with asymptomatic left ventricular dysfunction, are on the basis of the missed diagnosis. Today we hypotized that the uncorrected diagnosis could be, in several cases, the cause of sudden cardiac deaths. Consequently, would be important alert to this pathology especially paediatricians, sport doctors and cardiologists and we underline the necessity to have more adequate echocardiography machines used by trained operators.

References

1. Jenni R, Oechslin E, Schneider J, Attenhofer Jost C, Kaufmann PA. Echocardiographic and pathoanatomic characteristics of isolated left ventricular noncompaction: A step toward classification as a distinct cardiomyopathy. Heart 2001; 86: 666-671.
2. Elliot P, Andersson B, Arbustini E, et al. Classification of the cardiomyopathies: a position statement from the European Society of Cardiology Working Group on Myocardial and Pericardial Disease. Eur Heart J 2008; 29: 270-276.
3. Sasse-Klaassen S, Probst S, Gerull B, Oechslin E, Nurnberg P, Heuser A, Jenni R, Hennies HC, Thierfelder L. Novel Gene Locus for Autosomal Dominant Left Ventricular Noncompaction Maps to Chromosome 11p15. Circulation 2004; 109: 2720-2723.
4. Pignatelli RH, McMahon CJ, Dreyer WJ, Denfield SW, Price J, Belmont JW, Craigen WJ, Wu J, El Said H, Bezold LI, Clunie S, Fernbach S, Bowles NE, Towbin JA. Clinical characterization of left ventricular noncompaction in children: a relatively common form of cardiomyopathy. Circulation 2003; 108: 2672-2678.
5. Ritter M, Oechslin E, Sutsch G, Attenhofer C, Schneider J, Jenni R. Isolated noncompaction of the myocardium in adults. Mayo Clin Proc 1997; 72: 26-31.

Author Information

M Banci
1. Department of Cardiology, Valmontone Hospital, Rome, Italy

R Martinoli
2. Department of Biopathology and Imaging Diagnostics, University of Rome “Tor Vergata”, Rome, Italy

S Piccirilli
3. Department of Cardiology, University of Rome “Tor Vergata”, Rome, Italy

L Santucci
3. Department of Cardiology, University of Rome “Tor Vergata”, Rome, Italy

A Dofcaci
1. Department of Cardiology, Valmontone Hospital, Rome, Italy

F Papetti
1. Department of Cardiology, Valmontone Hospital, Rome, Italy

I Sansoni
4. Department of Imaging Diagnostic, University Campus Biomedical, Rome, Italy

P Saccucci
2. Department of Biopathology and Imaging Diagnostics, University of Rome “Tor Vergata”, Rome, Italy