Journal of the American College of Cardiology
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Published by Elsevier Science Inc.

Vol. 36, No. 2, 2000
ISSN 0735-1097/00/$20.00
PII S0735-1097(00)00755-5

Long-Term Follow-up of 34 Adults With Isolated Left Ventricular Noncompaction: A Distinct Cardiomyopathy With Poor Prognosis

Erwin N. Oechslin, MD, Christine H. Attenhofer Jost, MD, Jerry R. Rojas, MD, Philipp A. Kaufmann, MD, Rolf Jenni, MD, MSEE

Zurich, Switzerland

OBJECTIVES We sought to describe characteristics and outcome in adults with isolated ventricular noncompaction (IVNC).

BACKGROUND Isolated ventricular noncompaction is an unclassified cardiomyopathy due to intrauterine arrest of compaction of the loose interwoven meshwork. Knowledge regarding diagnosis, morbidity and prognosis is limited.

METHODS Echocardiographic criteria for IVNC include—in the absence of significant heart lesions—segmental thickening of the left ventricular myocardial wall consisting of two layers: a thin, compacted epicardial and an extremely thickened endocardial layer with prominent trabeculations and deep recesses. Thirty-four adults (age ≥16 years, 25 men) fulfilled the diagnostic criteria and were followed prospectively.

RESULTS At diagnosis, mean age was 42 ± 17 years, and 12 patients (35%) were in New York Heart Association class III/IV. Left ventricular end-diastolic diameter was 65 ± 12 mm and ejection fraction 33 ± 13%. Apex and/or midventricular segments of both the inferior and lateral wall were involved in >80% of patients. Follow-up was 44 ± 40 months. Major complications were heart failure in 18 patients (53%), thromboembolic events in 8 patients (24%) and ventricular tachycardias in 14 patients (41%). There were 12 deaths: sudden in six, end-stage heart failure in four and other causes in two patients. Four patients underwent heart transplantation. Automated cardioverter/defibrillators were implanted in four patients.

CONCLUSIONS Diagnosis of IVNC by echocardiography using strict criteria is feasible. Its mortality and morbidity are high, including heart failure, thrombo-embolic events and ventricular arrhythmias. Risk stratification includes heart failure therapy, oral anticoagulation, heart transplantation and implantation of an automated defibrillator/cardioverter. As IVNC is a distinct entity, its classification as a specific cardiomyopathy seems to be more appropriate. (J Am Coll Cardiol 2000;36:493–500) © 2000 by the American College of Cardiology

Noncompacted myocardium has been categorized as unclassified cardiomyopathy by the World Health Organization in the recently published report on definition and classification of cardiomyopathies (1). Noncompacted myocardium was previously described as persistent intramyocardial sinusoids. However, the latter are associated with congenital obstructive lesions of the left or right ventricular outflow tract, such as pulmonary atresia with intact ventricular septum (2–4). In these patients, regression of the embryogenic sinusoids is impaired during ontogenesis by ventricular pressure overload that results in deep recesses that communicate with both the ventricular cavity and the coronary artery system (3,5,6).

By contrast, isolated ventricular noncompaction (IVNC) is an idiopathic cardiomyopathy characterized by an altered structure of the myocardial wall as a result of intrauterine arrest of compaction of the myocardial fibers in the absence of any coexisting congenital lesion (7,8). There is continuity between the left ventricular (LV) cavity and the deep intratrabecular recesses that are filled with blood from the ventricular cavity without evidence of communication to the epicardial coronary artery system (9,10). Thus, the term "noncompaction" rather than "persistent sinusoids" is more appropriate for this condition (9,10). Since the original description, there have been only a few publications of case reports and small patient series describing predominantly a pediatric population (9,11–18). Despite an increasing awareness and interest in this anomaly, however, there is still little knowledge regarding diagnostic criteria, symptoms and prognosis of this rare and unique congenital disorder categorized as unclassified cardiomyopathy (1).

This study was designed to report clinical and echocardiographic characteristics and long-term outcome in the largest-ever described population of adults with IVNC.

METHODS

Diagnostic criteria. In the absence of other congenital or acquired heart disease, the typical echocardiographic image of noncompacted myocardium in IVNC is characterized by an altered structure of the LV myocardium with extremely thickened, hypokinetic segments consisting of two layers. There is thin, compacted myocardium on the epicardial side (epicardial layer) and thicker noncompacted myocardium on the endocardial side (endocardial layer), resulting in an extremely thickened ventricular wall. End-systolic thickness

From the Division of Echocardiography, University Hospital Zurich, Zurich, Switzerland.

Manuscript received October 28, 1999; revised manuscript received February 11, 2000, accepted March 30, 2000.

JACC Vol. 36, No. 2, 2000
August 2000:493–500

Abbreviations and Acronyms

ECG = electrocardiogram
IVNC = isolated ventricular noncompaction
LV = left ventricle or left ventricular
NYHA = New York Heart Association
N/C = noncompacted/compacted

of both the noncompacted epicardial and the compacted endocardial layer was measured at the site of maximal thickness to calculate the ratio of noncompacted/compacted (N/C). The measurements were taken at end-systole for best visualization of the two layers. The noncompacted endocardial layer has to be thicker than the compacted epicardial layer (ratio of N/C $\geq$2) and to consist of: 1) prominent and excessive trabeculations, and 2) deep recesses filled with blood from the ventricular cavity visualized on color Doppler imaging (Fig. 1). We have shown previously that diagnosis and localization of IVNC by echocardiography correspond to the necropsy specimens and to the findings of explanted hearts (10).

Study population. Between January 1984 and December 1998, 34 adults (age $\geq$16 years, 25 men) fulfilled the diagnostic criteria of IVNC (prevalence of 0.014% in patients referred to our echocardiography laboratory), and their data were prospectively entered into the database of our laboratory. Informed consent was given by all patients.

Data acquisition. Clinical assessment included a detailed medical history using standardized questionnaires. Medical records were reviewed. Sudden death was defined as occurring within 1 h of the patient's usual state of health or as unwitnessed death during sleep (19). All patients had a 12-lead resting electrocardiogram (ECG) and a complete two-dimensional Doppler echocardiographic exam as described below. Additionally, in 19 patients 24-h Holter monitoring was performed. Nonsustained ventricular tachycardia was defined as ventricular tachycardia of more than three premature ventricular contractions lasting up to 30 s. A ventricular run of more than 30 s was defined as sustained ventricular tachycardia.

Echocardiography. A complete two-dimensional and Doppler echocardiographic examination was performed in all patients according to the recommendations of the American Society of Echocardiography including two-dimensional guided M-mode measurements (20–22). Left ventricular ejection fraction was calculated using the biplane area length method (23).

The LV wall was divided into nine segments to describe the location of the noncompacted segments: the whole apex was one segment (apical segment); at the base and at the midventricular level, the LV was divided into four segments each (inferior, lateral, anterior and septal). The four midventricular segments were localized between the apical segment and the papillary muscles and the four basal segments between the papillary muscles and the base.

Diastolic function was assessed as previously described measuring the LV inflow pattern at the tip of the mitral valve leaflets and the pulmonary venous flow pattern in the right lower (or upper) pulmonary vein (24). Using previously published criteria, diastolic function was graded as follows: normal or abnormal relaxation, pseudonormal or restrictive pattern (24).

Follow-up. Prospective clinical follow-up was obtained in all 34 patients. Freedom from death and heart transplantation (nonsurvivors) were the combined end points of event-free survival. Heart transplant recipients were considered dead at the time of heart transplantation. Out of the 34 patients, serial echocardiographic data were available in 20 patients (59%). No serial echocardiography at our laboratory was obtained in 10 patients whose follow-up was less than two years (death, heart transplantation or recent diagnosis) and in four patients in whom follow-up echocardiography was performed by their private cardiologist after the diagnosis had been established.

Statistical analysis. Descriptive data for continuous variables are presented as means $\pm$ one standard deviation. Chi-square analysis or Fisher exact tests were used for nominal data. Continuous data were compared by the Mann-Whitney *U* test or the Wilcoxon rank-sum test. Probability of the event-free rate for the combined end point of death or heart transplantation was calculated by the Kaplan-Meier method of life table estimation. A p value of <0.05 was considered as statistically significant. Multivariate analysis was not performed, because of the small number of patients.

RESULTS

Clinical data. Clinical and electrocardiographic characteristics of the study population are presented in Table 1. The age range at the time of diagnosis was wide (16–71 years). Twenty-two patients (65%) were in New York Heart Association (NYHA) class I or II. Reasons for referral were: heart failure in 21 patients (62%); uncertain echocardiographic findings in four (12%); palpitations in two (6%); cardiomegaly, syncopal event, mitral regurgitation, myocarditis or pericarditis (one of each, 3%). One patient was referred to confirm the diagnosis of IVNC; one daughter of a patient was screened because of the familial occurrence risk and was diagnosed to have IVNC. Familial occurrence of IVNC was present in three families.

Echocardiographic data. Echocardiographic findings at the time of first presentation are summarized in Table 2. Left ventricular muscle mass cannot be calculated in patients with IVNC. An enlarged LV end-diastolic diameter ($\geq$60 mm) was present in 22 patients (67%), and reduced fractional shortening ($<$29%) in 27 of 33 patients (82%). Diastolic function could be assessed by echocardiography in 17 patients (50%); it was either not feasible (atrial fibrillation, tachycardia) or not yet done by Doppler echocardiography in the other patients.

Left ventricular thrombi were documented by echocardi-

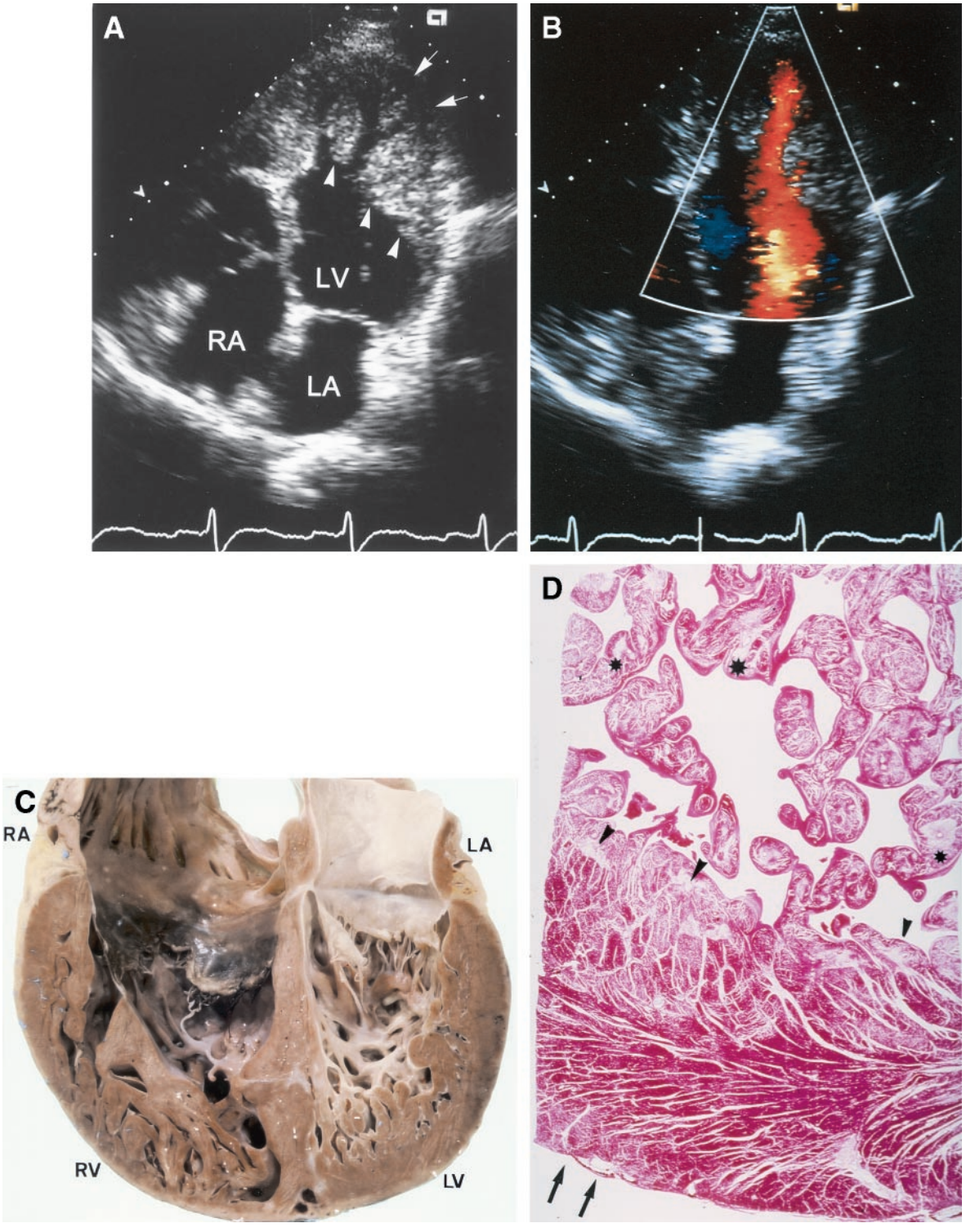

Figure 1. **(A)** Apical four-chamber view of a 66-year-old man. There is a thin epicardial layer **(thin arrows)** and an extremely thickened endocardial layer with prominent trabeculations and deep recesses **(arrowheads). (B)** Apical four-chamber view (end-diastolic still frame) of the same patient. There is blood flow from the ventricular cavity into the deep recesses visualized on color Doppler imaging. **(C)** Transsectional view from the anterior on the dorsal half of the heart of a 21-year-old man. There are both numerous trabeculations and deep recesses. Note marked fibroelastosis of the left ventricle. **(D)** Transmural, histologic section (hematoxylin and eosin stain). There are both an epicardial (compacted) layer **(arrows)** and an endocardial (noncompacted) layer. Note necrosis within the trabeculations **(asterisks)** as well as in the subendocardial area but not in the epicardial zone **(arrowheads).** LA = left atrium; LV = left ventricle; RA = right atrium; RV = right ventricle.

Table 1. Clinical and Electrocardiographic Characteristics in 34 Adults With IVNC at Initial Presentation

Male gender	25 patients (74%)
Age at diagnosis	42 ± 17 years
Chest pain	9 patients (26%)
Shortness of breath	27 patients (79%)
NYHA class I/II	22 patients (65%)
class III/IV	12 patients (35%)
Familial occurrence of IVNC	6 patients (18%)
Abnormal ECG	32 patients (94%)
Chronic atrial fibrillation	9 patients (26%)
Right bundle branch block	4 patients (12%)
Left bundle branch block	15 patients (44%)
Repolarization abnormalities (without block)	12 patients (35%)
Wolff Parkinson White syndrome	0 patient (0%)

ECG = electrocardiogram; IVNC = isolated ventricular noncompaction; NYHA = New York Heart Association.

ography in three patients, one of them with a history of mesenteric infarction.

Localization of noncompacted myocardial segments is shown in Figure 2. Most commonly, the apical and midventricular segments of both the inferior and lateral wall were affected (in more than 80% of patients). Involvement of the midventricular anterior wall and septum and the basal segments was much less frequent. Three and more segments were involved in 27 patients (79%). All noncompacted segments were hypokinetic. The normally compacted segments were occasionally also hypokinetic—despite normal wall thickness—which was reflected by the impaired fractional shortening in 27 patients (82%) consistent with basal hypokinesia. Left ventricular ejection fraction was <50% in 24 of 28 patients (86%).

Follow-up data. Duration of follow-up was 44 ± 39 months (range: 0.7–139). Mean age at the time of last follow-up (or before death or heart transplantation) was 46 ± 18 years (range: 16–71). Major complications are presented in Table 3. Heart failure (53%) requiring hospital admission was the most frequent event. Two patients presented in cardiogenic shock at the time of diagnosis; one of them died before transplantation could be attempted. The other patient was stabilized with medications, remaining in NYHA class II for eight years. Cardiogenic shock occurred in three other patients who could be medically stabilized using positive inotropic support in the intensive care unit.

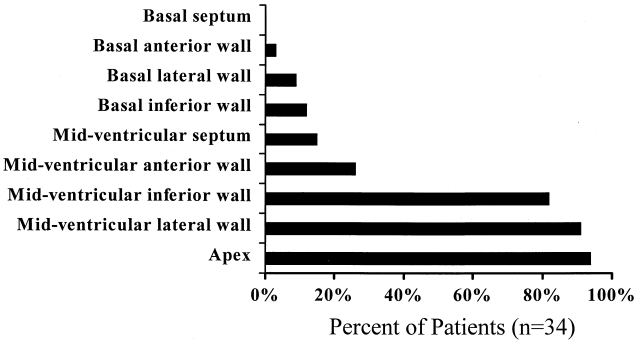

Figure 2. Distribution of segments affected by ventricular noncompaction.

Eleven thromboembolic events were observed in eight patients (24%). Eight systemic thromboembolic events occurred in seven patients (21%) including seven cerebrovascular accidents as the most common event in six patients (one episode of a stroke, six episodes of transient ischemic attacks); one patient suffered from a mesenteric infarction. Three episodes of pulmonary embolism were diagnosed in three patients. Two of the patients with pulmonary embolism also had cerebrovascular events or a mesenteric infarction, respectively.

Ventricular tachycardias were observed in 14 patients (41%): there were nonsustained ventricular tachycardias in 11 patients and sustained ventricular tachycardias in three patients. One patient with sustained ventricular tachycardia resistant to medical and electrical therapy died within 1 h after the onset of the arrhythmia. An automated internal cardioverter/defibrillator was implanted in four patients for sustained ventricular tachycardia (n = 2) or a presyncopal event and inducible ventricular tachycardia by electrophysiologic study (n = 2). One of these patients died from recurrent and refractory sustained ventricular tachycardia despite the implanted cardioverter/defibrillator.

In 20 patients with serial echocardiographic studies, there was no significant change of LV size or fractional shortening during follow-up (p = ns).

Table 2. Echocardiographic Data at Initial Presentation*

LVEDD	65 ± 13 mm	(normal: < 60 mm)
Fractional shortening	19 ± 10%	(normal: 29–46%)
Ejection fraction (n = 28)	33 ± 13%	(normal: ≥ 50%)
Left atrial size	4.4 ± 0.9 cm	(normal: 1.5–4.0 cm)
Thickness of basal septum	1.0 ± 0.2 cm	(normal: 0.6–1.2 cm)
Thickness of posterior wall	1.0 ± 0.4 cm	(normal: 0.6–1.2 cm)
Left ventricular thrombi	3 patients (9%)	
Diastolic function (n = 17)		
Impaired relaxation	6 patients (35.5%)	
Restrictive pattern	6 patients (35.5%)	
Pseudonormal	5 patients (29%)	
Normal	0 patient (0%)	

*Quantitative M-mode measurements were not available in one patient examined in the intensive care unit.
LVEDD = left ventricular end-diastolic diameter.

Table 3. Long-Term Follow-up Data

	Number of Patients
Heart failure requiring hospitalization	18 (53%)
Deaths	12 (35%)
Heart failure	4 (33%)
Sudden cardiac death	6 (50%)
Others*	2 (17%)
Heart transplantation	4 (12%)
Syncope	6 (18%)
NYHA class at last follow-up or before heart transplantation or death	
Class I/II	18 (53%)
Class III/IV	16 (47%)
Ventricular tachycardia	14 (41%)
Thromboembolic events	8 (24%)
Cerebrovascular accident	1 (3%)
Transient ischemic attack	6 (18%)
Mesenteric infarction	1 (3%)
Pulmonary embolism	3 (9%)

*Two other deaths included pulmonary embolism and arrhythmic, nonsudden death.
NYHA = New York Heart Association.

Mortality and heart transplantation. Eighteen patients (53%) were alive at last follow-up, 12 (35%) were dead, and 4 (12%) had undergone heart transplantation because of end stage heart failure (Table 3). Twelve patients died 42 ± 40 months (0.7–105) after diagnosis: sudden death occurred in six patients with stable NYHA class II (50% of all deaths), death from end stage heart failure in four (33%) and arrhythmic, non–sudden death in one (8.5%). Pulmonary embolism was the cause of death in another patient. Four patients underwent heart transplantation because of end stage myocardial failure at 3 to 33 months after diagnosis. The event-free rate for the combined end point of death or heart transplantation is shown in Figure 3. The probability of event-free survival (combined end point for death and heart transplantation) was 58% at five years. Mean age at death or heart transplantation was 45 ± 17 years (range 19–71).

Echocardiographic diagnosis was confirmed by pathologic-anatomic examination in nine patients: in five of them after death, in four of them after heart transplantation. Autopsy was not performed in seven deceased patients, because of refusal or death abroad.

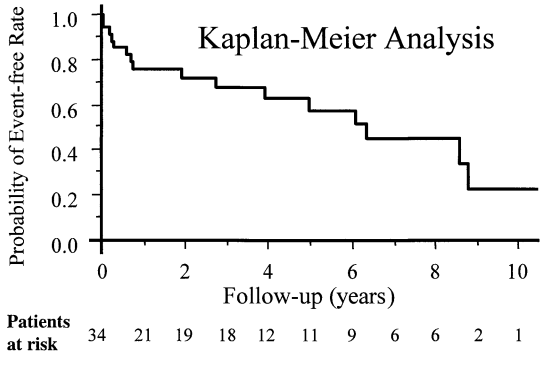

Figure 3. Probability of event-free rate for the combined end point of death or heart transplantation during follow-up.

Characteristics of survivors and nonsurvivors. The left ventricular end-diastolic diameter at the time of initial presentation was significantly larger in nonsurvivors (71 ± 9 mm) than it was in survivors (61 ± 12 mm; $p < 0.005$). New York Heart Association class III/IV (63% vs. 11%, $p < 0.005$), chronic atrial fibrillation (50% vs. 6%, $p < 0.005$) and bundle branch block (75% vs. 39%, $p < 0.045$) were more frequently present in nonsurvivors than in survivors. There was no significant difference between survivors and nonsurvivors regarding age at diagnosis, gender, heart failure requiring hospital admissions, fractional shortening and the presence of both embolic events and ventricular tachycardia.

DISCUSSION

Isolated ventricular noncompaction is a rare and unique congenital disorder categorized as unclassified cardiomyopathy in 1995 (1). Because of increasing awareness of this congenital anomaly after the initial publications, and advances in echocardiographic imaging, several more recent reports followed the first descriptions (7–18,25,26). Our series of 34 adults is the largest series so far. Clinical and echocardiographic characteristics and outcome during long-term follow-up are reported.

Clinical presentation. At presentation, the most common reasons for referral were heart failure and uncertain echocardiographic findings. In most patients (94%), the ECG was abnormal. Although embolic events were frequent complications, they were never the reason for referral. The age range at diagnosis or onset of heart failure varied widely. The only way to diagnose IVNC reliably was by echocardiography.

Diagnosis by echocardiography. Isolated ventricular noncompaction can be easily diagnosed by echocardiography if the echocardiographer is familiar with this congenital disorder and if clear cut diagnostic criteria are used. We have previously shown that the echocardiographic pattern allows both correct diagnosis and identification of segments involved in IVNC and there is excellent agreement with the necropsy findings (10). However, prominent LV trabeculations can be found in up to 68% of healthy hearts (27) and can be observed in hypertrophic hearts secondary to dilated, valvular or hypertensive cardiomyopathy. Thus, the differentiation between variants and IVNC may occasionally be challenging. However, the characteristic discriminating feature, which is crucial to diagnose IVNC, is the two layered myocardial wall structure with both a thin epicardial compacted zone and an extremely thickened endocardial noncompacted zone with deep recesses filled with blood from the ventricular cavity. Both the epicardial and endocardial layers of the myocardium in the noncompacted areas are perfused from the epicardial coronary arteries while the recesses are filled with blood directly from the LV cavity. We found the determination of the previously described X-to-Y ratio between the depth of intertrabecular recesses

Table 4. Characteristics of Isolated Ventricular Noncompaction in the Pediatric and Adult Population

	Chin et al.[9]	Ichida et al.[13]	Present Study
Number of patients	8 patients	27 patients	34 patients
Men	63%	56%	74%
Facial dysmorphism	38%	33%	0%
Age at diagnosis (median)	7 yrs	5 yrs	40 yrs
Follow-up (median)	Up to 5 yrs	Up to 17 yrs (6 yrs)	Up to 11 yrs (3 yrs)
Bundle branch block	25%*	15%	56%
Wolff Parkinson White syndrome	13%	15%	0%
Ventricular tachycardia	38%	0%	41%
Familial occurrence	50%	44%	18%
Heart failure symptoms	63%	30%	68%
Systemic embolic events	38%	0%	21%
Pulmonary embolism	0%	7%	9%
Ventricular thrombi	25%	0%	9%
Deaths	38%	7%	35%
Heart transplantation	0%	4%¶	12%
Localization of noncompacted segments			
Apex	Most prominent	100%	94%
Inferior wall		70%	94%
Lateral wall		41%	100%

*Left ventricular (intraventricular) conduction defects; ¶One patient was a candidate for heart transplantation.

relative to wall thickness helpful, but end-diastolic differentiation between noncompacted and compacted myocardium was often difficult (9). Therefore, we propose to identify first the segment with maximal wall thickness and then to assess the end-systolic thickness ratio between the noncompacted and the compacted layer. A ratio of noncompacted/compacted ≥ 2 is diagnostic for IVNC. In hearts with prominent trabeculations from other causes, the thickness ratio between trabeculated and normal zones never reaches the ratio of ≥ 2. By contrast to prominent trabeculations secondary to arterial hypertension or valvular disease, a segmental rather than a diffuse thickening or hypertrophy is present in patients with IVNC.

Another useful criterion is the location of the prominent trabeculations in patients with IVNC, which typically is apical, inferior and lateral and which is different from the prominent trabeculations found in normal or hypertrophied hearts. Prominent trabeculations as variants of normal hearts most frequently (85%) course from the free wall to the ventricular septum (27).

The right ventricular apex presents often with hypertrophic trabeculae separated by fissures, making the differentiation between variants of normal or pathologic patterns difficult. Thus, we do not attempt to diagnose right ventricular noncompaction as previously reported (10).

Morbidity and mortality. Major morbidity during long-term follow-up included heart failure, arrhythmias and thromboembolic events. Heart failure was caused by systolic and diastolic dysfunction. All our patients in whom diastolic function was assessed had diastolic dysfunction. Most of our patients (82%) also had diminished fractional shortening. Heart failure was the most common cause for hospital admission; it led to death and/or heart transplantation in eight patients (24%). The presence of noncompacted myocardium has, therefore, an impact on morbidity comparable with other cardiomyopathies.

Although depressed LV systolic function was found in 48% of children at initial presentation (13), clinically overt signs of heart failure were more frequent in our adult population. However, up to 89% of these children with a follow-up period longer than 10 years developed reduced LV function (13). In an older predominantly pediatric population, heart failure was as common as it was in our population (9). These observations suggest that IVNC present at birth is a progressive disorder resulting in systolic and diastolic heart failure during long-term follow-up, with a higher prevalence of heart failure in the adult than in the pediatric population. The cause of progressive myocardial failure has not yet been elucidated. The hypertrophic segments are perfused via the epicardial coronary arteries, which have no continuity to the deep recesses communicating with the LV cavity. Thickened endocardium and ischemic lesions in prominent trabeculae surrounded by the deep trabecular recesses were documented in histologic specimens, which may be caused by ischemia (10). Indeed, restricted myocardial perfusion in areas of IVNC was demonstrated by positron emission tomography (28). Hypothetically, both morphology and vasomotion of the coronary vessels feeding the hypertrophic segments may be abnormal with subsequent ischemia. Progressive ischemia and subsequent scar tissue may be an arrhythmogenic substrate for ventricular arrhythmias because the well-defined morphologic substrate of IVNC cannot be considered inherently arrhythmogenic.

Although IVNC in the basal segments was present only in a minority of the patients (Fig. 2), fractional shortening was reduced in 82% of our population as a consequence of progressive ventricular dilation with subsequent increase in

wall stress, subendocardial ischemia and fibrosis. However, these hypotheses need further investigation.

The high prevalence of thromboembolic events (24% of patients) was consistent with a previous report and was independent of LV size or function (9). The deep recesses may aggravate the risk of thrombus formation and be an additional factor for this serious complication. No systemic embolic events were found in the Japanese children, reflecting a younger and probably healthier population (13). In Chin's population, ventricular thrombi and systemic embolic events were even more common than they were in our population (9). Thus, we recommend oral anticoagulation for every patient in whom IVNC is diagnosed.

Long-term follow-up showed a high incidence of heart transplantation and death. Fifty percent of the patients died suddenly. Heart failure was the second most common cause of death. "Early" heart transplantation and "early" implantation of an automated cardioverter/defibrillator may be options for reducing the risk of premature death. We use this strategy for all of our patients with IVNC, especially for patients with NYHA class III or IV, atrial fibrillation and/or severely dilated LVs, which are more common in nonsurvivors. However, there are no data in this small population to support this strategy.

Comparison with pediatric population. Our data for adults and those of the two largest pediatric series so far published are summarized in Table 4 (9,13). Major discrepancies in the adult population were the lack of facial dysmorphism and the absence of Wolff Parkinson White syndrome. This may be explained by a different genetic background but the same morphologic appearance of the cardiac anomaly. In some chromosomal abnormalities (i.e. Xq28-linked noncompaction), prognosis is poor without survival until adulthood (11). Incomplete screening of the siblings may be a reason for the lower familial occurrence in our population. Complete bundle branch block was more common in adults. Other findings such as heart failure, ventricular tachycardias, embolic events or predominant involvement of the apical and inferior segments were comparable in adults and in an older pediatric population or adolescents. Although IVNC is a congenital disorder and present at birth, its clinical presentation may depend on the extent of noncompacted segments. Furthermore, the severity of this anomaly may progress during follow-up as a result of abnormal morphology and vasomotion of the vessels, with subsequent ischemia and progressive ventricular dilation as already hypothesized.

Study limitations. Our population consisted of patients referred to a tertiary care center because of severe heart failure or uncertain echocardiographic findings. Thus, there is selection bias in this population representing a highly selected group of mainly symptomatic patients. However, the true prevalence of this disorder in an unselected group consisting of both asymptomatic and symptomatic patients is not known, and the natural course of IVNC needs to be further elucidated.

Conclusions. This largest series of adults with IVNC to date confirms the high prevalence of heart failure, embolic events, ventricular arrhythmias and the poor prognosis in symptomatic patients. Anticoagulants should be administered independent of ventricular function to prevent embolic complications. Isolated ventricular noncompaction has a characteristic clinical, echocardiographic and pathologic pattern representing a pathologic anatomic entity distinguishable from other cardiomyopathies. Thus, IVNC should be classified as a specific cardiomyopathy comparable with other rare, distinct cardiomyopathies (e.g., arrhythmogenic right ventricular cardiomyopathy) to make the physician more familiar with this congenital disorder and its clear-cut diagnostic criteria.

Echocardiography is the method of choice to diagnose IVNC. There is a two-layered structure of the myocardial wall consisting of a thin compacted epicardial layer and a thick noncompacted endocardial layer with prominent trabeculations and deep recesses. Because of the risk of familial occurrence, first-degree relatives should be screened by echocardiography to identify asymptomatic patients. As physicians are becoming more aware of this rare disorder, previously missed patients may be diagnosed, and the true prevalence and natural course of IVNC will be better elucidated and understood in the future.

Acknowledgements

The authors are indebted to Jakob Schneider, MD, and Peter Vogt, MD, Department of Pathology, University Hospital, Zurich, for providing the pathologic-anatomic specimens.

Reprint requests and correspondence: Dr. Rolf Jenni, Division of Cardiology, University Hospital Zurich, Raemistrasse 100, CH-8091 Zurich, Switzerland. E-mail: karjer@usz.unizh.ch.

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