# Family screening in black patients with isolated left ventricular non-compaction: the Chris Hani Baragwanath experience

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## Abstract

**Background:** Isolated left ventricular non-compaction (ILVNC), dilated cardiomyopathy (DCMO) and hypertrophic cardiomyopathy (HCM) are diseases that may be present in family members of patients with ILVNC. The primary aim of this study was to identify the prevalence and spectrum of cardiomyopathy in first-degree relatives of patients with ILVNC. A secondary aim was to compare a strategy of clinical screening, utilising only a clinical assessment and electrocardiogram (ECG), compared to one that included echocardiography for screening of family members of patients with ILVNC.

**Methods:** Eighty-three close relatives of 38 unrelated patients from the ILVNC clinic at the Chris Hani Baragwanath Hospital underwent a detailed clinical history, physical examination, ECG and echocardiogram.

**Results:** Echocardiographic screening revealed unexplained left ventricular (LV) dysfunction in 10 (12.05%) relatives. Nine out of the 10 individuals satisfied the criteria for diagnosis of DCMO. No cases of HCM or LVNC were identified. A strategy of clinical assessment and ECG had a sensitivity of 76% and a specificity of 42% versus the gold standard of echocardiographic screening.

**Conclusion:** Echocardiographic screening detected DCMO in 10.8% of subjects. A strategy of clinical screening that included electrocardiography was sub-optimal as a screening strategy compared to echocardiographic screening.

**Keywords:** family screening, left ventricular non-compaction

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Isolated left ventricular non-compaction (ILVNC) is a primary myocardial disorder that is presumed to be genetic, according to the American Heart Association.[1] The term isolated is used when there is no evidence of accompanying congenital, valvular or associated cardiomyopathy disorders. The prevalence of ILVNC in sub-Saharan Africa is not clearly defined. Peters *et al.* found in their series that 6.9% of patients in a tertiary cardiomyopathy clinic had ILVNC.[2] Clinical findings are variable, ranging from patients with asymptomatic disease to symptomatic patients who develop congestive cardiac failure, arrhythmias, thromboembolic events and sudden cardiac death.[3]

ILVNC, dilated cardiomyopathy and hypertrophic cardiomyopathy are diseases that may be present in family members of patients with ILVNC. Hence, early identification of family members may offer the opportunity for early detection of complications of LVNC (such as arrhythmias and thrombi), dilated cardiomyopathy and hypertrophic cardiomyopathy. These individuals may require intervention with appropriate therapy, which may translate into a potential benefit.

The aim of this study was to identify, during family screening, the prevalence and spectrum of cardiomyopathy in family members of patients with ILVNC. A secondary aim was to determine the value of clinical screening, which utilises only a clinical assessment and electrocardiogram (ECG), compared with screening using echocardiography.

## Methods

This retrospective study was undertaken at the Left Ventricular Non-compaction Clinic, Chris Hani Baragwanath Hospital, on existing clinical and echocardiographic records of first-degree relatives of known patients with ILVNC. From January 2014 until July 2016, first-degree relatives of patients diagnosed and followed up for ILVNC were invited to undergo family screening.

After providing voluntary informed consent, family members underwent a detailed clinical history. The family history was considered abnormal if it was positive for non-ischaemic heart failure, cardiomyopathy, documented supraventricular or ventricular arrhythmias, or pacemaker/implantable cardioverter–defibrillator placement. Thereafter, they underwent a clinical examination followed by a resting ECG. The ECG was analysed to measure the heart rate, and P-R, QRS and Q-T intervals. The ECG was considered abnormal if it showed pathological Q waves (> 40 ms or > 25% R waves in ≥ two leads), abnormal axis, left ventricular hypertrophy, complete bundle branch block, or non-specific intraventricular conduction delay.

A transthoracic echocardiogram was performed on all subjects who were screened according to a standardised protocol by three experienced, accredited sonographers using a Philips IE 33 system, equipped with a standard S5-1 transducer. The images were obtained according to a standardised protocol.

The data were transferred and analysed offline using Xcelera workstation (Philips). All linear and volumetric chamber

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| Table 1. Diagnosis of ILVNC, DCMO and HCMO | |
|---|---|
| ILVNC | A combination of the echocardiographic criteria of Oechslin *et al.*[3] and Stöllberger[7] were used for the diagnosis of ILVNC in this study. These criteria have previously demonstrated the ability to distinguish normal individuals from subjects with ILVNC in a sub-Saharan African population. A diagnosis of ILVNC was made when all four of the following criteria were present:<br>• A ratio of non-compacted to compacted myocardium > 2 when measured at end-systole<br>• The presence of more than three prominent trabeculations in the left ventricular apex that did not originate from the septum<br>• Deep intertrabecular recesses that filled with blood from the ventricular cavity as visualised on colour Doppler ultrasound<br>• No evidence of congenital or acquired heart disease |
| DCMO | Left ventricular or biventricular systolic dysfunction and dilatation that is not explained by abnormal loading conditions or coronary artery disease. LVEF < 45% associated with left ventricular dilatation[8] |
| Hypokinetic non-dilated cardiomyopathy | Left ventricular or biventricular global systolic dysfunction without dilatation (defined as LVEF < 45%), not explained by abnormal loading conditions or coronary artery disease[8] |
| HCM | The presence of left ventricular wall thickness ≥ 15 mm in one or more left ventricular myocardial segments with no other haemodynamics or metabolic cause[9] |
| ILVNC: isolated left ventricular non-compaction, DCMO; dilated cardiomyopathy, HCM: hypertrophic cardiomyopathy. | |

measurements were performed according to the American Society of Echocardiography (ASE) chamber guidelines[4] by two accredited readers, S Nel and F Peters. Left ventricular end-diastolic volumes, end-systolic volumes and left ventricular ejection fraction (LVEF) were measured using the Simpson method. Measurements relating to left ventricular diastolic function were performed as per the ASE guidelines on diastolic function and included pulse-wave Doppler at the mitral tips and tissue Doppler of both medial and lateral mitral annuli.[5] Measurements relating to the right ventricle were based on ASE guidelines on the right ventricle.[6] Echocardiography was used to specifically diagnose ILVNC, dilated cardiomyopathy (DCMO) and hypertrophic cardiomyopathy (HCM) based on predefined criteria (Table 1).

Continuous variables are summarised as the mean ± standard deviation as appropriate. Categorical variables are presented as frequencies and percentages. Sensitivity, specificity, positive and negative predictive values, and the likelihood ratio for a positive or negative test for ECG against echocardiography as the gold standard were calculated with their 95% confidence interval.

## Results

The baseline characteristics of all family members screened are summarised in Table 2. A total of 83 close relatives of 38 unrelated patients with ILVNC accepted our invitation for screening. Pre-existing hypertension was found in 11 (13.2%) of the screened members. With regard to these 11 family members, two had a history of previous cerebrovascular accident. The majority of family members who were screened were asymptomatic. However, three were symptomatic with two having New York Heart Association (NYHA) class II dyspnoea and one being in NYHA class III.

A total of 83 ECGs were performed, with 61 (74.5%) subjects having a normal ECG (Table 3). Abnormal findings were observed in 22 subjects (26.5%). In 16 of the 22 subjects, left ventricular hypertrophy (LVH) was detected (72.7%). Only one of these subjects with LVH had pre-existing hypertension.

Echocardiographic screening revealed unexplained left ventricular dysfunction in 10 (12.05%) of the cohort of relatives screened (Table 4). Of the 10 participants with unexplained LV dysfunction, one had pre-existing hypertension, and nine had no known pre-existing cardiovascular abnormalities. The remaining nine individuals satisfied the criteria for the diagnosis of dilated cardiomyopathy. None of the participants met any of the criteria for ILVNC, HCM or hypokinetic DCMO. Six of these subjects diagnosed with DCMO were asymptomatic whereas two were in NYHA class II and one was in NYHA class III. Three of the nine individuals diagnosed with DCMO were from the same family.

A comparison between a strategy of clinical and ECG screening only versus echocardiographic findings (Table 5) revealed that 61 (73.5%) subjects had a normal clinical evaluation and a normal ECG. Within this group, abnormal echocardiograms were found in seven (11.5%) subjects. The echocardiographic abnormalities found were six subjects had DCMO and one had unexplained LV dysfunction but did not meet the criteria for DCMO, HCM or LVNC.

| Table 2. Baseline characteristics of screened non-compaction cardiomyopathy relatives | |
|---|---|
| Patients (*n*) | 83 |
| Age at presentation (years) | 30.7 ± 15.3 |
| Females, *n* (%) | 46 (55.2) |
| Pre-existing hypertension, *n* (%) | 11 (13.2) |
| Systolic blood pressure (mmHg) | 127 ± 21 |
| Diastolic blood pressure (mmHg) | 78 ± 14 |
| Heart rate (beats/min) | 76 ± 15 |
| Connection, *n* (%) | |
| Parent | 9 (10.8) |
| Sibling | 29 (35.0) |
| Child | 41 (49.4) |
| Other* | 4 (4.8) |
| NYHA class, *n* (%) | |
| I | 80 (95.4) |
| II | 2 (3.4) |
| III | 1 (1.2) |
| *Other: niece, nephew, aunt or uncle. NYHA: New York Heart Association. | |

| Table 3. ECG characteristics of screened non-compaction cardiomyopathy relatives | |
|---|---|
| Patients (*n*) | 83 |
| Sinus rhythm, *n* (%) | 83 (100) |
| Heart rate (beats/min) | 76 ± 15 |
| Abnormal axis, *n* (%) | 3 (3.6) |
| Left | 2 (66.7) |
| Right | 1 (33.3) |
| Bundle branch block | 1 |
| LBBB | 0 |
| RBBB | 1 |
| Left ventricular hypertrophy, *n* (%) (Sokolow–Lyon criteria) | 16 (19.3) |
| LBBB: left bundle branch block, RBBB: right bundle branch block. | |

**Table 4. Echocardiographic characteristics of screened non-compaction relatives**

| | |
|---|---|
| Variable (*n*) | 83 |
| LVEDD (mm) | 44.1 ± 5.5 |
| LVESD (mm) | 29.9 ± 5.4 |
| Ejection fraction (%) | 59.8 ± 6.2 |
| End-diastolic volume (ml/m$^2$) | 88.4 ± 25.9 |
| End-systolic volume (ml/m$^2$) | 36.3 ± 14.6 |
| IVS (mm) | 8.9 ± 2.0 |
| Relative wall thickness (mm) | 0.4 ± 0.1 |
| Posterior wall thickness (mm) | 8.5 ± 2.0 |
| E wave (cm/s) | 87.3 ± 22.8 |
| A wave (cm/s) | 66.2 ± 23.8 |
| E/A (ratio) | 1.6 ± 0.5 |

LVEDD: leeft ventricular end-diastolic diameter, LVESD: left ventricular end-systolic diameter, IVS: interventricular septal diameter.

**Table 5. Echocardiographic findings if a strategy of clinical examination and ECG analysis were used**

| | |
|---|---|
| Subjects with normal clinical exam and a normal ECG, *n* (%) | 61 (73.5) |
| Normal echo | 54 (88.5) |
| Abnormal echo | 7 (11.5) |
| Subjects with a normal clinical exam and an abnormal ECG, *n* (%) | 22 (26.5) |
| Normal echo | 17 (77.3) |
| Abnormal echo | 5 (22.7) |

**Table 6. Sensitivity and specificity of clinical and ECG screening**

| | |
|---|---|
| Sensitivity (%) | 76 |
| Specificity (%) | 41.7 |
| Positive predictive value (%) | 88.5 |
| Negative predictive value (%) | 22.7 |
| The likelihood ratio for a positive test | 1.31 |
| The likelihood ratio for a negative test | 0.57 |

Twenty-two (26.5%) of the subjects had a normal clinical evaluation but an abnormal ECG. Within this group, abnormal echocardiograms were found in five (22.7%). The echocardiographic abnormalities found were three subjects had DCMO, one had hypertensive heart disease with diastolic dysfunction, and one had an incidental finding of a pericardial effusion. The sensitivity and specificity of clinical and ECG screening versus the defined gold standard of echocardiographic diagnosis of cardiomyopathy (ILVNC/DCMO/HCM) are depicted in Table 6.

## Discussion

The main findings of this study are that family screening detected the phenotype of dilated cardiomyopathy in 10.8% of subjects with two-thirds of these individuals being asymptomatic. No cases of ILVNC or HCM were detected. The second major finding was that a screening strategy that utilised clinical evaluation and an ECG was moderately sensitive in detecting cardiomyopathy in comparison with cardiological screening, which utilised echocardiography.

ILVNC is presumed to be a genetic disorder,[1] and consequently, family screening has been advocated to detect pathology in asymptomatic individuals. In this study, family screening identified the phenotype of dilated cardiomyopathy in nine (10.8%) previously undiagnosed individuals, of whom six were asymptomatic, with three of these individuals belonging to the same family. Despite some individuals having prominent trabeculation, none of the individuals with the phenotype of DCMO satisfied the criteria used in our study for the diagnosis of ILVNC. Furthermore, no cases of HCM were identified.

Our findings differ from other family screening studies, which found ILVNC in between 18 and 50% of subjects,[10-15] DCMO in between 12 and 15%[16,17] and HCM in between 3 and 7% of subjects.[16,17] These differences may be attributed to variations in the diagnostic screening strategy employed, the population studied, imaging techniques and criteria used, and referral bias relating to this study.

The interplay between LVNC and DCMO is an important consideration for the clinician. If the index case is presumed to be ILVNC with or without a dilated cardiomyopathy phenotype, family screening may reveal a DCMO phenotype without ILVNC in screened relatives.[18-20] This most often arises in families where there are sarcomeric gene mutations. The converse finding of relatives with ILVNC phenotype discovered during family screening where the index cases are DCMO has also been described.[19] Hence the discovery of a *de novo* case of ILVNC with either a dilated cardiomyopathy or a DCMO phenotype may result in the discovery of diverse genotype–phenotype manifestations when family screening is performed.

Several studies have highlighted the differences in detection of affected family members based on the screening strategy employed.[16,20] Echocardiographic screening has the advantage of identifying the phenotype of ILVNC, DCMO or HCM in individuals who are screened irrespective of whether they have any cardiac symptoms. It has been suggested that up to 63% of individuals with a phenotypic abnormality on routine screening are asymptomatic. When only family history was used without echocardiographic screening, 44% of individuals would not have their phenotypic abnormality identified.[16] Furthermore, since genetic abnormality has only been detected in 50% of cardiologically screened confirmed cases of ILVNC,[16] it implies that cardiological screening allows for more robust identification of abnormality.

A major disadvantage not employing accompanying genetic screening is that individuals with non-penetrance/reduced penetrance may not be identified. Identifying individuals with non-penetrance may require recurrent cardiac screening of affected carriers, although the results of such a strategy have not been adequately studied. Similarly, it is unknown whether repeat cardiac screening is required in unaffected individuals from families where the genetic abnormality is unknown.

This study comprised a cohort of adults over the age of 18 years. Several screening studies have included screening children as well. A study in Australia on 314 children over a 10-year period found ILVNC in 9.2%, HCM in 25.5% and DCMO in 58.6% of patients.[23] In a recent publication, which represents the largest screening study conducted to date, van Waning *et al.* found that mutations may be more common in children.[17] Therefore by excluding children, we may have underestimated the prevalence of abnormality in our study.

A second issue relates to ethnicity since our cohort comprised only individuals who were African. Ethnic differences may result in various gene abnormalities and phenotypic expression related to left ventricular remodelling. Therefore it may be that African family members of individuals with either sporadic or

genetic ILVNC may manifest more commonly with the dilated cardiomyopathy phenotype. However, no definitive conclusion in this regard may be drawn until further work is conducted, since to our knowledge no other screening studies have been conducted in African families of subjects with ILVNC.

Echocardiography is the most commonly used technique to diagnose ILVNC, as it is widely available, feasible and non-invasive. However, echocardiography has several limitations, which can lend itself to over- or under-diagnosis of ILVNC.[24,25] Echocardiography is highly dependent on the operator's technical skill to acquire suitable images and requires proper interpretation of data received. There is also concern about the reproducibility of current diagnostic criteria, which has demonstrated poor inter-observer agreement.[26] Given these limitations, cardiac magnetic resonance imaging (MRI) has a far superior spatial resolution, less operator dependence and higher contrast in the myocardium, which can provide better delineation of the trabeculations.

A cohort study done by Diwardkar *et al*. showed how echocardiography failed to detect ILVNC in patients diagnosed with it on MRI.[24] In our study, cardiac MRI was not utilised, and this may have improved the diagnosis of individuals with ILVNC in subjects who were difficult to image or where echocardiography missed the pathology.

One of the major findings of this study was that utilising a strategy of clinical evaluation and ECG had a sensitivity of 76% and a specificity of 42% versus the gold standard of echocardiographic screening. In resource-deprived settings, such a strategy may be attractive if it is used as the initial screening strategy and followed by echocardiographic screening with or without genetic screening. For such a strategy to be successful, the initial screening strategy must ideally have a very high sensitivity, which often implies a lesser degree of specificity. Our findings highlight the failure of using the ECG in addition to clinical evaluation as an initial screening strategy due to its relatively modest sensitivity. Furthermore, the role of a 12-lead ECG as a screening tool alone for ILVNC is debatable, as there are no specific ECG patterns to diagnose ILVNC.

In a cohort study done by Steffel *et al*.,[27] the most common findings on initial diagnosis of ILVNC were intraventricular conduction delay, voltage signs of LVH and repolarisation abnormalities. A completely normal ECG was present in only 13% of patients. These abnormal ECG signs can be found in normal African individuals. Lohrmann *et al*.[28] showed that early repolarisation patterns occurred in 53.2% of subjects. LVH occurred in 13% and bundle branch blocks in 0.5% of normal black adults with echocardiographically normal hearts. Therefore using a strategy of clinical evaluation and an ECG in this study was inferior since the sensitivity and specificity of results were sub-optimal for screening.

Limitations of this study were that it was a retrospective study with a small study population and therefore the external validity of our secondary aim was limited. Not all eligible family members were screened. Genetic testing and cardiac MRI were not performed.

## Conclusion

Echocardiographic screening detected DCMO in 10.8% of subjects whereas no cases of ILVNC or HCM were identified. A strategy of clinical screening that included electrocardiography was sub-optimal as a screening strategy compared to echocardiographic screening in this study.

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## Landmark study highlights importance of cholesterol monitoring of young adults

Adults as young as 25 years, not only older people, need to know their 'bad cholesterol' [non-high-density lipoprotein cholesterol (non-HDL-C)] levels so they can change their lifestyle or take drugs to protect themselves against heart attacks and strokes in later life. The landmark study, involving data from nearly 400 000 people in 19 countries, establishes for the first time that levels of non-HDL-C or 'bad cholesterol' in the blood, are closely linked to the risk of heart disease across the entire life course.

The research could lead to many younger people taking statins to lower their cholesterol levels. At the moment GPs prescribe the cholesterol-lowering drugs mostly to people in middle age. The authors said it was important to know your 'bad cholesterol' level from young adulthood; it gave you the chance to lower the level through exercise, a healthier diet, or by taking statins.

'We need to start it early,' said Stefan Blankenberg, a professor in Hamburg, Germany, who was part of the multinational cardiovascular risk consortium that carried out the modelling study. He said would he like to see new guidance for doctors. 'We should at least put into the guidelines that non-HDL-C determination should be an obligation. At a very young age – 25 to 30 years. You need to know it.'

He added: 'In German schools we have large anti-smoking programmes. We persuade populations not to smoke. We have no programme to let people know about cholesterol. The first thing I would do is establish a cholesterol knowledge programme.' For young adults the first remedy for high non-HDL-C would be exercise and losing extra weight, followed by eating a healthier diet, said Blankenberg.

Colin Baigent, director of the MRC Population Health Research Unit, at the University of Oxford, is quoted in the report as saying: 'This is an important paper because it shows what could be achieved if, starting early in their 40s, healthy people were to start taking a statin so that their bad cholesterol is halved for the rest of their lives.

'Of course, despite the fact that statins are safe and well tolerated, many healthy people would be reluctant to take a statin from early middle age. But the striking findings of this study show that a policy of recommending such treatment might be a long-term investment that leads to a substantial improvement in the health of older people in the years to come.'

*Source:* Medical Brief 2020