Choc cardiogénique fulminant révélant une non-compaction ventriculaire gauche chez un nourrisson de 3 mois

Fulminant Cardiogenic Shock as the Initial Presentation of Left Ventricular Non Compaction in a 3 Month Old Infant

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Résumé

La non-compaction du ventricule gauche (NCVG) est une cardiomyopathie congénitale rare, résultant d'un arrêt du développement myocardique, caractérisée par des trabéculations importantes et une altération de la fonction ventriculaire. Nous rapportons le cas d'un nourrisson de sexe masculin âgé de 3 mois, sans antécédents médicaux, admis pour détresse respiratoire aiguë et collapsus circulatoire. À son admission, il était en choc cardiogénique nécessitant une ventilation mécanique et un traitement vasopresseur. Les examens biologiques ont révélé une leucopénie sévère, une anémie microcytaire et une élévation marquée du taux de troponine, sans syndrome inflammatoire. L'échocardiographie a mis en évidence une dysfonction systolique sévère du ventricule gauche (fraction d'éjection à 20 %) avec un aspect de non-compaction de la paroi latérale. L'imagerie par résonance magnétique cardiaque a confirmé le diagnostic. L'état du patient s'est initialement amélioré sous dobutamine et traitement de l'insuffisance cardiaque, permettant un sevrage de la ventilation et des vasopresseurs. Cependant, son état s'est ensuite dégradé et il est décédé malgré une prise en charge intensive maximale.

Ce cas souligne la présentation néonatale fulminante de la NCVG et son mauvais pronostic lorsqu'elle se manifeste par un choc cardiogénique.

Mots-clés

Non-compaction du ventricule gauche, cardiomyopathie, choc cardiogénique, insuffisance cardiaque, échocardiographie, IRM cardiaque.

Summary

Left ventricular non compaction (LVNC) is a rare congenital cardiomyopathy resulting from arrested myocardial development, characterized by prominent trabeculations and impaired left ventricular function. We report the case of a 3 month old male infant with no prior medical history who presented with acute respiratory distress and circulatory collapse. On admission, he was in cardiogenic shock requiring mechanical ventilation and vasoactive support. Laboratory investigations revealed severe leukopenia, microcytic anemia, and markedly elevated troponin levels, without evidence of inflammatory syndrome. Echocardiography demonstrated severe left ventricular systolic dysfunction (ejection fraction 20%) with a non compaction pattern involving the lateral wall. Cardiac magnetic resonance imaging confirmed the diagnosis. The patient initially improved with dobutamine and heart failure therapy, enabling successful weaning from ventilation and vasopressors. However, he subsequently deteriorated and died despite maximal intensive care.

This case underscores the fulminant neonatal presentation of left ventricular non compaction and its poor prognosis when manifesting as cardiogenic shock.

Keywords

Left ventricular non-compaction, cardiomyopathy, cardiogenic shock, heart failure, echocardiography, cardiac MRI

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INTRODUCTION

Left ventricular non compaction (LVNC) is an uncommon cardiomyopathy characterized by excessive trabeculations and deep intertrabecular recesses, resulting from arrested myocardial compaction during embryogenesis. Although increasingly recognized across all age groups, it is rare in infancy. Clinical presentation ranges from incidental detection to severe heart failure, arrhythmias, thromboembolic events, and sudden cardiac death (1–3). In pediatric populations, LVNC is particularly severe when presenting early in life and is often associated with ventricular dysfunction and poor outcomes (4,5). Diagnosis relies primarily on echocardiography and cardiac magnetic resonance imaging, which demonstrate a two layered myocardium with a non compacted to compacted ratio above established diagnostic thresholds (6). We report a case of fulminant cardiogenic shock leading to the diagnosis of LVNC in a previously healthy 3 month old infant.

CASE PRESENTATION

A 3-month-old male infant with no significant past medical history was admitted to the Pediatric Intensive Care Unit for acute respiratory distress and hemodynamic collapse. On admission, he was in severe shock with cyanosis, tachypnea, prolonged capillary refill time (4 seconds), marked tachycardia (200 beats per minute), and severe hypotension (50/25 mmHg). Abdominal examination revealed no hepatomegaly. The presentation suggested a low cardiac output state.

Emergency management included immediate endotracheal intubation with mechanical ventilation, fluid resuscitation, and initiation of norepinephrine for refractory hypotension. Electrocardiography showed T-wave inversion in lateral leads, indicating possible myocardial involvement. Laboratory investigations revealed severe leukopenia (2,070/mm^3), microcytic hypochromic anemia, and markedly elevated troponin levels (2,200 ng/L), without evidence of systemic inflammation, suggesting acute myocardial dysfunction rather than infectious shock.

Transthoracic echocardiography demonstrated severe left ventricular systolic dysfunction (ejection fraction 20%), a prominent non-compaction pattern of the lateral wall, and moderate mitral regurgitation. No congenital structural heart disease was identified. Cardiac magnetic resonance imaging confirmed the diagnosis of left ventricular non-compaction.

Given the cardiac origin of shock, norepinephrine was discontinued and inotropic support with dobutamine (20 µg/kg/min) was initiated, resulting in rapid hemodynamic improvement. The patient was progressively weaned from mechanical ventilation and vasoactive agents. Guideline-directed medical therapy for heart failure was introduced, including spironolactone, bisoprolol, furosemide, and sacubitril/valsartan. Despite initial stabilization, sudden clinical deterioration occurred a few days later, and the patient died despite maximal intensive care management.

DISCUSSION

Left ventricular non-compaction (LVNC) is a rare, heterogeneous cardiomyopathy resulting from arrested myocardial compaction during embryogenesis, producing a two-layered myocardium with a thin compacted epicardial layer and a prominent non-compacted endocardial layer with excessive trabeculations and deep recesses. (1-3) While some individuals remain asymptomatic for years, pediatric cases—particularly those in infancy—are often associated with severe ventricular dysfunction and poor outcomes (4,5).

Clinical presentation ranges from incidental imaging findings to progressive heart failure, arrhythmias, thromboembolic events, and sudden cardiac death (1-3). In neonates and infants, severe systolic dysfunction may develop early due to both structural abnormalities and limited myocardial reserve (4). Our case illustrates an extreme phenotype with fulminant cardiogenic shock at 3 months of age, underscoring the aggressive nature of early-onset LVNC.

Pathophysiologically, LVNC reflects incomplete myocardial compaction between the 5th and 8th weeks of gestation (1,2), leading to impaired mechanics, subendocardial hypoperfusion, and arrhythmia susceptibility. In our patient, global systolic dysfunction (ejection fraction 20%) and markedly elevated troponin suggested severe myocardial stress rather than ischemic injury. The absence of inflammatory markers and imaging findings supported a primary cardiomyopathy over myocarditis.

Electrocardiographic changes in LVNC are nonspecific (3); in this case, lateral T-wave inversion indicated myocardial involvement but was not diagnostic. Imaging was central: echocardiography revealed the characteristic two-layered myocardium, and cardiac MRI confirmed LVNC and excluded other structural anomalies.

Transthoracic echocardiography is the first-line diagnostic

tool for left ventricular noncompaction (LVNC), typically revealing a two-layered myocardium with prominent trabeculations and deep recesses, most often involving the apical and lateral segments of the left ventricle (6,9). Diagnostic criteria vary, and interobserver variability remains a limitation. Cardiac magnetic resonance imaging (MRI) has become essential for confirmation, providing superior spatial resolution and enabling quantification of the non-compacted to compacted myocardial ratio (6,9). In our patient, MRI definitively confirmed LVNC and excluded other structural abnormalities.

Genetic factors are increasingly recognized in LVNC, with mutations identified in sarcomeric, cytoskeletal, and mitochondrial genes (7,8). These overlaps with other cardiomyopathies support the concept that LVNC may represent a morphological phenotype rather than a distinct disease entity in some cases. Although genetic testing was not performed in our patient, the very early and severe presentation suggests a potentially pathogenic mutation, with implications for family screening and genetic counseling.

Management of LVNC is largely supportive, based on standard heart failure therapy including diuretics, beta-blockers, and modulation of the renin–angiotensin–aldosterone system (10). Evidence in infants is limited, and treatment is extrapolated from other forms of pediatric dilated cardiomyopathy. In our case, transient improvement followed inotropic support with dobutamine and initiation of guideline-directed therapy, indicating partial reversibility of the hemodynamic component.

Despite initial stabilization, the clinical course was rapidly fatal, underscoring the unpredictable evolution of severe infantile LVNC and the limited impact of medical therapy in advanced stages. Early-onset LVNC with severe ventricular dysfunction is associated with high mortality and may progress to end-stage heart failure despite optimal treatment. (5,11).

Heart transplantation remains the only definitive option in refractory cases; however, rapid deterioration in infants may preclude timely evaluation and listing, as occurred in our patient. This highlights the importance of early referral to specialized pediatric heart failure and transplant centers when severe LVNC is suspected.

This case emphasizes the need to differentiate LVNC from other causes of acute heart failure in infancy, including myocarditis, metabolic cardiomyopathies, and structural congenital heart disease. A multimodal diagnostic approach combining clinical assessment, echocardiography, cardiac MRI, and, when feasible, genetic testing is essential for accurate diagnosis and prognostication.

CONCLUSION

Ce cas souligne l'importance d'une évaluation exhaustive devant une embolie pulmonaire du sujet jeune, en particulier lorsque l'imagerie révèle une masse intracavitaire. L'IRM cardiaque permet une caractérisation précise, essentielle pour éviter les erreurs diagnostiques. Le déficit combiné en Protéine C et Protéine S, bien que rare, doit être évoqué devant une thrombose atypique et non provoquée.

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