Severe foetal hypertrophic cardiomyopathy evolving to left ventricular non-compaction

Pedro Betrián Blasco*, Dimpna Calila Albert Brotós, Queralt Ferrer Mendiña, Ferrán Rosés Noguer, and Gemma Giralt García

Pediatric Cardiology, ‘Valle de Hebrón’ Hospital, Paseo de Valle de Hebrón 119-129, CP 08035 Barcelona, Spain

Received 31 May 2010; accepted after revision 1 July 2010; online publish-ahead-of-print 28 July 2010

A 29-week-old male foetus was diagnosed by foetal echocardiography with severe hypertrophic cardiomyopathy with systolic dysfunction and generalized oedema, undergoing a Caesarean section at 33 weeks. Mechanical ventilation and milrinone infusion were required during the first week. Systolic function and output parameters improved progressively. Metabolic and infectious screenings were negative. At the follow-up, during the first year of life, hypertrophy regressed, posterior right auricular hypertrophy evolved to a mass with cysts, and left ventricular myocardium developed trabeculations accomplishing non-compaction criteria. Recently, mutations in genes previously implicated in the pathogenesis of hypertrophic cardiomyopathy have been identified in patients with left ventricular non-compaction without hypertrophy. This report suggests that these cardiomyopathies may have a similar genetic origin, and can co-exist in the same patient.

Keywords
Morphogenesis • Hypertrophy • Echocardiography • Non-compaction

A 29-week-old male foetus was diagnosed by foetal echocardiography with severe hypertrophic cardiomyopathy with important atrial involvement and moderate systolic dysfunction, undergoing a Caesarean section at 33 weeks. Familiar history was anodyne. Neonatal echocardiography confirmed diagnosis (left ventricular posterior wall, 5.5 mm; septum, 7.5 mm; ejection fraction, 45%). Mechanical ventilation and milrinone infusion were required during the first week. Systolic function and output parameters improved progressively (Figure 1, Supplementary data, Videos 1 and 2). Metabolic and infectious screenings were negative.

At the follow-up, during the first year of life, the patient developed a mild psychomotor delay with axial hypotony and central respiratory insufficiency requiring home mechanical ventilation. Echocardiographic controls in this first year showed progressive regression of hypertrophy, except at the lateral right auricular wall, where persisted (Figure 2, Supplementary data, Video 3). At the same time, left ventricular myocardium developed trabeculations accomplishing left ventricular non-compaction criteria,\textsuperscript{1,2} with no changes in ventricular function (Figure 3, Supplementary data, Video 4). Familial echocardiographic screening has been negative.

Recently, mutations in genes encoding sarcomeric proteins, which previously have been implicated in the pathogenesis of

\* Corresponding author: Avda de Les Marines 52 4ª 1º, Sant Cugat del Vallés, Barcelona, Spain. Tel: +34 699 163 749; fax: +34 934 893 039. Email: pedrobetrian@yahoo.es

Published on behalf of the European Society of Cardiology. All rights reserved. © The Author 2010. For permissions please email: journals.permissions@oxfordjournals.org.

Figure 1
Newborn, echocardiography: four-chamber view. Severe biventricular hypertrophy with important atrial involvement.
hypertrophic cardiomyopathy, have been identified in patients with left ventricular non-compaction but without hypertrophic cardiomyopathy (MYH7, ACTC, TNNT2).3,4 Initially assumed to be congenital, left ventricular non-compaction in the meantime has been shown to develop during life-time in several cases.5 Literature has reported the relationship of left ventricular non-compaction and neuromuscular disorders,4 the co-existence of left ventricular non-compaction and hypertrophic cardiomyopathy in the same family,5 and patient.6 Our report shows the overlapping of these two entities and their evolutive and dynamic character. We do not know why hypertrophic cardiomyopathy appeared first and evolved to left ventricular non-compaction, perhaps the presence of systolic foetal heart failure, could have been the trigger to hypertrophy, and after the birth, when systolic function improved, progressive regression of the hypertrophy and evolution to left ventricular non-compaction was observed.

Supplementary data
Supplementary data are available at European Journal of Echocardiography online.

References