Prenatal echographic recognition of hypertrophic cardiomyopathy leading to heart transplantation in the newborn

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A healthy, non-diabetic 27-year-old woman attended a routine foetal echography during the 32nd week of an uneventful first pregnancy. Growth of the (female) foetus was normal for the gestational phase and no general malformation was observed. However, the echocardiographic evaluation (Panel A) showed massive left ventricular (LV) hypertrophy (end-diastolic thickness of the interventricular septum, 13 mm; left posterior wall, 22 mm); the right-ventricular free wall was also hypertrophic. These findings were confirmed at birth (Panel B) by echocardiographic demonstration of massive cardiac hypertrophy, with a 20 mm maximal LV wall thickness uncorrected for body surface area. A systolic gradient >30 mmHg was detected in both ventricular outflow tracts. LV ejection fraction was 80%. ECG (not shown) was diagnostic for biventricular hypertrophy, with deep inferior and anterolateral Q-waves. The PR interval was normal. Metabolic diseases were excluded based on the results of comprehensive blood and urine analyses. Screening for the beta-myosin heavy chain, cardiac myosin binding protein C, and cardiac troponin I gene mutations was negative. Both parents had normal physical examinations, ECG, and echocardiography. No family history of heart disease could be traced. From the first hours after birth, the baby girl presented severe congestive heart failure, which was unresponsive to aggressive pharmacological treatment. Heart transplantation was therefore performed at the age of 2 months, and 20 months later the girl is currently in good health. The gross specimen of the explanted heart (Panel C) showed massive hypertrophy of both ventricles, with extremely diminutive cavities. Histological examination showed widespread disarray of the cardiac muscle cells (Panel D) and ruled out any storage or inflammatory disease.

Hypertrophic cardiomyopathy (not secondary to metabolic disorders) can occasionally be congenital. In utero recognition of this disease allows timely planning of clinical management, including possible need for heart transplantation.

Panel A. Foetal echography at 32 weeks shows massive cardiac hypertrophy involving the right-ventricular free wall, interventricular septum, and LV posterior wall.

Panel B. Postnatal echocardiography shows cardiac morphology similar to that in foetal echocardiography. Panels C and D. Gross and histopathological features from the explanted heart show right and left ventricular hypertrophy (interventricular septum, 19 mm; LV posterolateral wall, 20 mm; right-ventricular free wall, 12 mm) and cardiac muscle cell disorganization (myocyte disarray).

AO, aorta; Liv, liver; LL, left lung; LV, left ventricle; RV, right ventricle; RL, right lung; S, spine.

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