

## Variant in a very preterm newborn: a case report neonatal onset of inherited myocardial hypertrophy due to rare MYBPC3

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**Background.** Hypertrophic cardiomyopathy (HCM) is an inherited myocardial disease present in one in 500 of the general population. It represents the most frequent cause of sudden cardiac death of young people. Inherited HCM in the newborn is rare. Genetic testing is necessary to discriminate transient forms of neonatal HCM, which have been related to gestational diabetes and exposition to prenatal or postnatal corticosteroids in preterm infants.

**Case presentation.** A male infant was born after 31 weeks of gestation to a 45-year-old primigravida, primipara woman who had a medical history of an inherited HCM caused by MYBPC3 mutation. Pregnancy was obtained through homologous blastocyst transfer. Fetal echocardiography performed at 24 weeks of gestation resulted normal. The infant was delivered via cesarean section for non-reassuring fetal monitoring.

Initial neonatal resuscitation in the delivery room included intubation, positive pressure ventilation, and oxygen supplementation. Apgar scores resulted 2, 4, and 6 at 1, 5, and 10 minutes, respectively. Afterwards, a transthoracic echocardiography revealed moderate left-ventricular wall thickness, suggestive of an HCM.

**Conclusions.** Because of the family history of heart disease and due to the persistency of HCM in the postnatal period, genetic testing by next-generation sequencing with a comprehensive cardiomyopathy panel was performed. A heterozygous single-nucleotide missense familial variant was found in the MYBPC3 gene (c.3713T > C p. Leu1238Pro). This anomaly is likely pathogenetic of inherited HCM and is a rare variant which is not detected in the ExAC population database (> 60,000 samples) according to the Atlas of Cardiac Genetic Variation.