# A case of fetal hypertrophic cardiomyopathy in second trimester

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## Objective

Hypertrophic cardiomyopathy (HCM) is an autosomal dominant disease. In pediatric settings, pure HCM has an estimated incidence of 4. 7 per million children. Despite HCM being often sub-clinical, sudden cardiac death may occur as the first manifestation of the disease. Cardiomyopathies are very rare in fetuses and seem to be associated to poor outcome. To the best of our knowledge, only isolated case reports and small case series have been reported. We present a case of pre-natal diagnosis of HCM in second trimester.

## Methods

Case report.

## Results

A 32 year-old healthy women, G5P1 was referred to our prenatal diagnosis unit at 23 weeks of gestation due to a symmetric thickness of fetal cardiac biventricular wall. Familial history of hypertrophic cardiomyopathy has just been diagnosed during the current pregnancy. The genetic study of the affected family members (mother and cousin of the pregnant women) detected the variant c. 559A>G (p. Asn187Asp) in heterozygosity for MYH7 gene. This missense variant is not described in literature. The fetal echocardiogram (23 weeks) revealed thickened biventricular wall and intra-ventricular septum, measuring about 4. 5 mm and 3 mm, respectively. Good cardiac function and no obstacle in ventricular outflow were observed. Fetal karyotype revealed a normal female karyotype. The family variant in heterozygosity for MYH7 gene was detected in fetus by DNA sequencing. The presence of the family variant in the pregnant women was unknown and her echocardiogram was found to be normal. According to the Portuguese law and after genetic counseling, the patient decided for termination of pregnancy. After the reunion of the ethical committee the pregnancy was terminated in the 24 week of gestation and the anatomopathological examination confirmed the concentric biventricular hypertrophy.

## Conclusion

Prenatal diagnosis of HCM is associated with complex medical and psychological implications because HCM expression is highly variable and its evolution is difficult to predict. Even though, early echocardiographic findings appear to be associated with more adverse perinatal outcomes.