

Aortic dilatation and miscarriages as a main presentation of FLNA mutation in a Croatian family: a case report

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KEYWORDS: FLNA mutation, miscarriage, aortic dilatation, aortic dissection.

CITATION: *Cardiol Croat.* 2021;16(5-6):188. | <https://doi.org/10.15836/ccar2021.188>

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Introduction: The FLNA gene provides instructions for producing protein filamin A. It is found on the X chromosome and has X linked inheritance. The dysfunction of this gene is associated with congenital malformation of the cerebral cortex, cardiac abnormalities, thoracic aneurism and joint hypermobility.¹⁻⁴

Case report: We present a family with heterozygous pathogenic variant of FLNA. Disease was discovered during workup of older daughter's miscarriages. She had a double miscarriage in the first trimester. She knew for mild mitral and aortic regurgitation from youth. From family history: mother had two miscarriages and two successful deliveries, pulmonary hypertension, coronary artery disease and percutaneous coronary intervention at the age of 56, grandmother from mother side had one successful delivery and three miscarriages, died at the age of 64 from diabetic coma. Sister has moderate aortic regurgitation, dilatation of ascending aorta (42mm), one miscarriage. Father has dilatation of ascending aorta. In 2020, echocardiography revealed dilatation of the ascending aorta (43mm) with mild central aortic regurgitation, and a trace of mitral regurgitation. The cardiologist recommended CT aortography and genetic testing. On CT aortography aorta was measured at a maximum of 46mm (**Figure 1**). Genetic testing identified one pathogenic variant in FLNA. After this discovery, genetic testing was performed on all family members (**Figure 2**), and mutation was identified in the mother and sister. During this workup, the younger sister found out that she was pregnant. Soon, the mother suddenly died at the age of 60. Autopsy revealed a dissection of the thoracic aorta. With this finding, the younger sister went from category three to category four in classification of maternal cardiovascular risk score and was advised to abort. At the time of writing this paper she is 10 weeks pregnant and, knowing all the risks, does not want an abortion.



FIGURE 1. CT scan, dilatation of the ascending aorta.

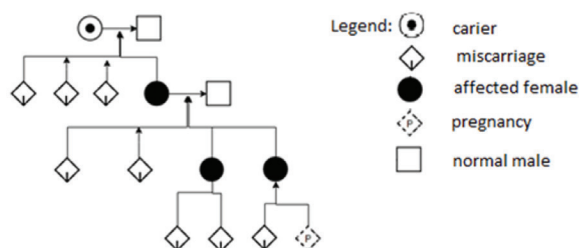


FIGURE 2. Genetic tree.

RECEIVED:
March 28, 2021

ACCEPTED:
April 2, 2021



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