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

Marija Tomac Stojmenović^{1*},

Vlatka Rešković Lukšić²,

©lrena Ivanac Vranešić².

Velena Radošević², Tamara Žigman²,

Maja Hrabak Paar²,

DJadranka Šeparović Hanževački²

¹Psychiatric Hospital Rab, Rab, Croatia

²University of Zagreb School of Medicine, University Hospital Centre Zagreb, Zagreb, Croatia **KEYWORDS:** FLNA mutation, miscarriage, aortic dilatation, aortic dissection.

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*ADDRESS FOR CORRESPONDENCE: Marija Tomac Stojmenović, Psihijatrijska bolnica Rab, Kampor 224, HR-51280 Rab, Croatia. / Phone: +385-95-910-2378 / E-mail: marija.tomac.mts@gmail.com

ORCID: Vlatka Rešković Lukšić, https://orcid.org/0000-0002-4721-3236 • Irena Ivanac Vranešić, https://orcid.org/0000-0002-6910-9720 Maja Hrabak Paar, https://orcid.org/0000-0002-0390-8466 • Jadranka Šeparović Hanževački, https://orcid.org/0000-0002-3437-6407

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 suc-

cessful 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.

of writing this paper she is 10 weeks pregnant and, knowing all the risks, want an abortion.

Conclusion: In young women with dilatation of ascending aorta, connective tissue disease should be considered. Today we have possibility for family screening, so we can discover pathogenic mutation in families. In FLNA mutation, safe size of ascending aorta dilatation is smaller than in healthy population, which should be considered during pregnancy planning. In case of pregnancy multidisciplinary approach is mandatory.



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

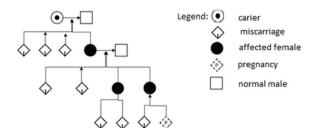


FIGURE 2. Genetic tree.

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