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LETTER

Identification of a Missense Mutation in the FLNC Gene from a Chinese Family with Restrictive Cardiomyopathy [Letter]

Hasta Handayani Idrus[1,2]

[1]Department of Biomedical Sciences, Postgraduate School, Hasanuddin University (UNHAS), Makassar, Indonesia; [2]Center for Biomedical Research, Research Organization for Health, National Research and Innovation Agency (BRIN), Cibinong Science Center, Cibinong-Bogor, West Java, Indonesia

Correspondence: Hasta Handayani Idrus, Department of Biomedical Sciences, Postgraduate School, Hasanuddin University (UNHAS), Perintis Kemerdekaan Road KM.05, Makassar, South Sulawesi, Indonesia, Email hastahandayani99@gmail.com; hast006@brin.go.id

Dear editor

We have read the paper by Jiangtao Dong et al on Identification of a Missense Mutation in the FLNC Gene from a Chinese Family with Restrictive Cardiomyopathy.[1] We congratulate all authors who have provided new information regarding the whole-exome sequencing (WES) method, which has been proven effective in finding genes that cause genetic diseases, one of which is Restrictive Cardiomyopathy (RCM). Cardiomyopathy is a heterogeneous myocardial disease associated with electrical dysfunction in the heart muscle, resulting in hypertrophy or dilation of the ventricles of the heart.[2] Pathogenic mutations in the FLNC gene can be an early genetic screening in patients or families of patients with suspected restrictive cardiomyopathy; this is important for assessing the risk of carrier patients and aims to intervene early so that it does not become a more severe case.[3]

The study conducted by Jiangtao Dong et al took blood samples from the probandus and his healthy parents and then used the whole-exome sequencing and Sanger sequencing methods to identify possible pathogenic genes.[1] The procedure used in this study was correct, it should be noted that whole-exome sequencing can not only be the basis for molecular diagnosis but can also predict the development and clinical therapy of cardiomyopathy patients, in addition the FLNC gene contains the most P/LP variants so that FLNC truncation can cause severe clinical symptoms in cardiomyopathy patients.[4]

The study conducted by Jiangtao Dong et al showed that the results of missense mutations in FLNC were detected in the proband through whole-exome sequencing and Sanger sequencing, although not found in the parents, the mutation can damage and change the normal structure and function of FLNC, which contributes to the occurrence of restrictive cardiomyopathy.[1] However, the results can be influenced by the administration of heart medication such as furosemide, spironolactone, and metoprolol, which can improve the proband's heart function and reduce the symptoms experienced. Therefore, early detection of intervention from restrictive cardiomyopathy is needed earlier in order to block the gene.[3]

In conclusion, we agree that FLNC missense mutations known to be pathogenic in hypertrophic cardiomyopathy can be detected early with whole-exome sequencing and Sanger sequencing methods, which can be recommended examinations;[1] this can help in gene load analysis, variant classification, and genotype–phenotype correlation analysis that can help more appropriate therapy in restrictive cardiomyopathy patients.[5] As additional information, the FLNC gene can also be a screening for other heart diseases such as dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy, and other heart phenotypes such as arrhythmias without detectable structural abnormalities, congenital heart disease, and noncompaction cardiomyopathy.[3]

Disclosure

Author reports no other conflict of interest in this communication.

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