Clinical Genetics

AB081. Variable major phenotypes in familial Marfan syndrome in Indonesia: a case report

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Background: Mutations in *FBN1* gene can cause Marfan syndrome (MFS), in which 25% of cases are familial type. Affected persons show varying patterns of organ system involvements including ocular, skeletal, cardiovascular, pulmonary, dural and cutaneous manifestation. Although genotype-phenotype correlations have been described in some cases. The majority of MFS patients still expressed a wide phenotypic variability, even within family with the same mutation.

Methods: Family members of an index case with MFS were examined, followed by echocardiography and X-ray. *FBN1* gene mutation screening was performed using multiplex ligation-dependent probe amplification (MLPA) and denaturing high performance liquid chromatography (DHPLC). DNA sequencing was performed to confirm *FBN1* mutation for any case with positive screening.

Results: In six family members, *FBN1* mutation c.1924G>T, designated as p.Gly642Ter, were found. Despite of harboring the same mutation, each of affected family members had different major phenotypes, ranging from aortic dissection and dilatation, heart valve abnormalities, spontaneous pneumothorax and severe kyphoscoliosis.

Conclusions: Identification of MFS patients should lead to further investigation in other family members. The intrafamilial variation warrants a thorough clinical investigation for individual management. DNA analysis is important test for establishing the diagnosis of MFS and for predicting the recurrence risk.

Keywords: Marfan syndrome (MFS); *FBN1* mutations; phenotypes; Indonesian

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