

Marfan Syndrome in Semarang: report of two cases

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Abstract

Marfan Syndrome is a heritable connective tissue disorders, mostly caused by mutations in fibrillin-1 gene. In major cases, this syndrome is inherit in autosomal dominant manner, with ~25% cases are caused by de novo mutations. Affected persons showed varying patterns of organ involvement including the ocular, skeletal, cardiovascular, pulmonary, dura and skin, in which aortic dissection become the most common cause of mortality. High variability in phenotypic expressions between and within families were also observed in this syndrome.

Two cases of Marfan Syndrome in whom mutation analysis in *FBN1* gene have been performed, showed two novel mutations. The first case was a 7-years old boy with a de novo missense mutation in exon 28 of *FBN1* gene, c.3545G>A [p.C1182T]. The patient presented with lens dislocation, aortic dilatation, mitral valve prolapsed, pectus carinatum and dolichostenomelia. The second case was a familial Marfan Syndrome. The proband was a 32-years old man with a nonsense mutation in exon 15 of *FBN1* gene, c.1924G>T [p.Gly642X]. He presented with tall stature, increased arm span and height ratio, bilateral high myopia, arachnodactily, positive thumb signs and wrist signs, joint laxity of articulation genu, history of spontaneous pneumothorax, and finally passed away because of aortic dissection. His mother, two sisters and brother were clinically Marfan Syndrome, and will be further described.

The variability in phenotypes and disorders severity should be better considered. The presence of Marfan Syndrome affected person in a family should be followed by further investigation in other family members to conclude the inheritance manner. DNA analysis is important for diagnostic establishment and knowing the recurrence risk in the next generation. Early recognition in affected status will lead to early prevention to complications that may follow. Comprehensive management including genetic counseling thus needed for Marfan Syndrome patients and their family members.

Key words : *Marfan Syndrome, FBN1 mutation, Semarang*