Newborn Screening, Inborn Errors of Metabolism

AB092. A novel variant c.1506G>C at exon 9 of iduronate 2-sulfatase gene in mucopolysaccharidosis type II Indonesian child

Yulia Ariani, Rizky Priambodo, Cut Nurul Hafifah, Damayanti Rusli Sjarif

Department of Medical Biology, Faculty of Medicine Universitas Indonesia, Senen, Kota Jakarta Pusat, Indonesia

Background: Mucopolysaccharidosis type II is one of the lysosomal storage diseases caused by the absence or malfunctioning of iduronate 2-sulfatase (IDS) enzyme in the lysosome due to alteration of *IDS* gene. This case report aimed to demonstrate our first MPS type II patient which tested for genetic analysis using Sanger sequencing method.
Methods: Pathogenic variants are frequently reported being located at exon 9, especially in Asian population. Due to limited resources we first tried to do Sanger sequencing at exon 9. A pair of primers reported in one study from Taiwan are being used to amplify exon 9. Using Sanger sequencing, this exon is sequenced.
Results: A 3-year-old boy, diagnosed as MPS type II based on a very low I2S enzyme activity. This patient was first brought to Department of Neurology due to speech delay and facial changes since 1.5 years old. There was no motor delay, but mother noticed stiffness of both hands. Patient is the first child from Indonesia non-consanguineous parents. He was born spontaneously with unremarkable history. He has normal growth and development until 1 year old. Physical examination showed coarse facies, frontal bossing, enlarged adenoid and joint arthropathy. Supporting examination revealed acetabular shape of cervical vertebrae, trivial mitral regurgitation, and mild hypomyelination. One single-nucleotide alteration variant in exon 9 was found. This alteration has never been reported elsewhere. This novel variant (c.1506G>C) leads to the alteration of amino acid from tryptophan to cysteine (p.Trp502Cys).
Conclusions: A novel variant (c.1506G>C) has been found in this case study. This might be a candidate disease-causing mutation which affects the normal structure and function of I2S enzyme. Further steps should be done to test this. Finding this variant in control group would be the first step to be done.

Keywords: Mucopolysaccharidosis type II; iduronate 2-sulfatase enzyme; iduronate 2-sulfatase gene; Indonesian

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