Analisis mutasi ekson 2 dan ekson 5--6 gen PTS pada penderita defisiensi enzim 6-pyruvoyl tetrahydropterin synthase (PTPS) di Indonesia = Mutation analysis for exon 2 and exon 5--6 of PTS gene on 6-pyruvoyl tetrahydropterin synthase (PTPS) deficiency patient in Indonesia

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Abstrak

Defisiensi enzim 6-pyruvoyl tetrahydropterin synthase PTPS menyebabkan terjadinya hambatan dalam proses biosintesis tetrahydrobiopterin BH4 yang merupakan kofaktor berbagai jenis enzim, termasuk phenylalanine hydroxylase PAH. Enzim PAH tidak dapat diaktivasi tanpa adanya senyawa BH4, sehingga menyebabkan timbulnya penyakit langka yang disebut dengan hyperphenylalaninemia HPA. Penelitian ini dilakukan untuk menganalisis mutasi yang terjadi pada ekson 2 dan 5--6 gen PTS di Indonesia. Analisis mutasi dilakukan pada 3 penderita defisiensi enzim PTPS dan 50 individu normal asal Indonesia.
Tahapan analisis mutasi pada penelitian ini diawali dengan melakukan desain primer spesifik dan penentuan suhu annealing optimal dengan menggunakan PCR gradien. Sequencing kemudian dilakukan dengan metode automated Sanger sequencing yang dilanjutkan dengan analisis hasil sequencing untuk mengetahui mutasi yang terdapat pada ekson 2 dan 5--6 gen PTS di Indonesia. Hasil yang didapatkan pada penelitian ini yaitu, tiga mutasi novel pada ekson 2 yaitu c.123G>A, c.127T>G, serta c.155A>T, serta tidak ditemukan mutasi pada ekson 5--6.
<hr>Deficiency of 6 pyruvoyl tetrahydropterin synthase PTPS enzyme can interrupt biosynthesis of tetrahydrobiopterin BH4 , which is a cofactor of various enzymes, including phenylalanine hydroxylase PAH. The PAH enzyme can not be activated in the absence of BH4 compounds, leading to the occurrence of a rare disease called hyperphenylalaninemia HPA. This study was conducted to analyze the mutations that occurred in exon 2 and 5 6 of the PTS gene in Indonesia. The mutation analysis was performed on 3 patients with PTPS enzyme deficiency and 50 normal individuals from Indonesia.
Stages of mutation analysis in this study is began by performing specific primer design and optimal annealing temperature determination using PCR gradient. Sequencing is then performed by automated Sanger sequencing method followed by sequencing analysis to find out the mutations found in exon 2 and 5 6 of the PTS gene in Indonesia. The results obtained in this study are three novel mutations in exon 2 which are c.123G A, c.127T G, and c.155A T, and no mutations found in exon 5 6.