Analisis mutasi gen PTS pada ekson 1 dan ekson 3--4 dari penderita defisiensi enzim 6-pyruvoyl tetrahydropterin synthase (PTPS)di Indonesia = Mutation analysis of PTS gene exon 1 and exon 3-4 from 6-pyruvoyl tetrahydropterin synthase PTPS enzyme deficiency patient in Indonesia

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Abstrak

Defisiensi enzim 6-pyruvoyl tetrahydropterin synthase PTPS merupakan salah satu penyakit yang disebabkan oleh aktivitas enzim PTPS. Enzim PTPS memiliki peran dalam proses biosintesis tetrahydrobiopterin BH4. Defisiensi enzim PTPS menyebabkan gangguan untuk proses biosintesis BH4 sehingga, tidak dapat mengubah senyawa fenilalanin menjadi senyawa tirosin, disebut hyperphenylalanemia HPA. Defisiensi enzim PTPS terjadi akibat adanya mutasi pada gen PTS, dapat mengubah struktur dan fungsi dari asam amino yang dihasilkan.
Penelitian tersebut bertujuan untuk menganalisis mutasi gen PTS ekson 1 dan ekson 3--4 yang terjadi pada penderita defisiensi enzim PTPS di Indonesia. Sampel DNA yang digunakan adalah hasil isolasi DNA darah pada tiga penderita defisiensi enzim PTPS di Indonesia dan 50 individu normal 25 laki-laki dan 25 perempuan. Sekuens gen PTS pada ekson 1 dan ekson 3--4 dari sampel tersebut diamplifikasi menggunakan metode PCR.
Hasil dari proses PCR divisualisasikan menggunakan elektroforesis gel, kemudian disekuensing menggunakan metode automated Sanger sequencing. Hasil yang didapat dalam peneltian ini adalah tidak ditemukan mutasi pada penderita defisiensi enzim PTPS di ekson 1 dan ekson 3--4, namun ditemukan adanya mutasi di intron 4, yang bersifat novel yaitu IVS4 5T>C dan IVS4 6G>T.
<hr>The 6 pyruvoyl tetrahydropterin synthase PTPS enzyme deficiency is one of the diseases caused by the activity of enzyme PTPS. The PTPS enzyme has a role in the biosynthesis of tetrahydrobiopterin BH4, when the enzyme is disturbed, it can not convert phenylalanine into a tyrosine, called hyperphenylalanemia HPA. The PTPS enzyme deficiency caused a disruption BH4 biosynthesis so, can not convert phenylalanine into a tyrosine, called hyperphenylalanemia HPA. PTPS enzyme deficiency occurs due mutations in the PTS gene, can changed the structure and function of the amino acids produced.
This aim of this research are for analyze the mutation of exon 1 and exon 3 4 in PTS gene of patients with PTPS enzyme deficiency in Indonesia. DNA samples were extracted from the blood three patients with PTPS enzyme deficiency in Indonesia and 50 normal individuals 25 male and 25 female. The DNA samples were amplified using PCR method.
The results of the PCR process were visualized using gel electrophoresis, then were sequenced using the automated Sanger sequencing method. This study figure of that were no mutations found in patients with PTPS enzyme deficiency in exon 1 and exon 3 4, but two novel mutation found in intron 4 which are IVS4 5T C and IVS4 6G T.