Analisis mutasi gen iduronat 2-sulfatase ekson 4 dan 7 pada penderita mukopolisakaridosis tipe II di Indonesia = Mutation analysis of iduronate 2-sulfatase gene exon 4 and 7 on mucopolysaccharidosis type II patients in Indonesia

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Abstrak

Mukopolisakaridosis tipe II MPS II merupakan suatu sindrom yang disebabkan oleh defisiensi enzim iduronat 2-sulfatase I2S yang dikode oleh gen iduronat 2-sulfatase IDS. Mutasi pada gen IDS dapat menyebabkan perubahan struktur dan fungsi dari enzim I2S yang dihasilkan. Penelitian ini dilakukan untuk mengetahui dan menganalisis mutasi gen iduronat 2-sulfatase IDS ekson 4 dan 7 pada penderita MPS II di Indonesia. Sampel DNA diekstraksi dari darah 9 individu penderita MPS II dan 50 individu normal 25 laki-laki dan 25 perempuan. Sekuens gen IDS ekson 4 dan 7 dari sampel-sampel tersebut diamplifikasi menggunakan metode PCR.
Hasil dari proses PCR divisualisasi menggunakan Agarose Gel Electrophoresis AGE, kemudian disekuensing menggunakan metode automated sequencing. Hasil penelitian menunjukkan adanya mutasi delesi c.435_440delTACCGA yang merupakan varian likely pathogenic dan mutasi silent c.489G>A yang merupakan varian likely benign pada ekson 4, serta satu mutasi missense yang merupakan varian pathogenic pada ekson 7, yaitu c.998 C>T.
<hr>Mucopolysaccharidosis type II MPS II is a syndrome which is caused by deficiency of iduronate 2 sulfatase enzyme, coded by iduronate 2 sulfatase IDS gene. Mutation in IDS gene can alter structure and function of the resulting I2S enzyme. This study was conducted to analyze IDS gene mutations of exon 4 and 7 in mucopolysaccharidosis type II patients in Indonesia. DNA samples were extracted from the blood of 9 MPS II patients males and 50 normal individuals which consists of 25 males and 25 females. The sequence of IDS gene exon 4 and 7 from those samples were amplified using PCR method.
PCR results were visualized using Agarose Gel Electrophoresis AGE, and were sequenced using automated sequencing. The results showed one deletion c.435 440delTACCGA which is classified as likely pathogenic variant and one silent mutation c.489G A which is a likely benign variant on exon 4, and one missense mutation of pathogenic variant on exon 7, c.998 C T.