# Analisis mutasi pada ekson 6 dan 8 gen iduronat 2-sulfatase pada pasien mukopolisakaridosis tipe II di Indonesia = Mutation analysis in exon 6 and 8 of the iduronat 2-sulfatase in mucopolysaccharidosis type II patients in Indonesia

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Abstrak

Mukopolisakaridosis II MPS II, atau sindrom Hunter, adalah penyakit resesif terpaut-X langka yang disebabkan oleh gangguan penyimpanan lisosomal akibat defisiensi enzim lisosomal iduronate-2-sulfatase IDS. Gen IDS penting dalam proses degradasi lisosomal dermatan sulfat dan heparan sulfat, karena defisiensi gen IDS akan mengarah pada akumulasi Glikosaminoglikans GAG tersebut. Analisis spesifik ekson pada ekson 6 dan ekson 8 gen IDS dilakukan pada penderita MPS II di Rumah Sakit Umum Pusat Nasional Dr. Cipto Mangunkusumo, Jakarta, Indonesia. Dalam penelitian ini, sampel dari penderita MPS II dan sampel normal dianalisis menggunakan PCR dan metode sekuensing.
Hasil yang diperoleh menunjukkan mutasi-mutasi novel pada dua penderita MPS II di Indonesia. Satu mutasi insersi sepanjang 14 pb c.792_793insCCCCTGTGGCCTAC ditemukan pada ekson 6 dari gen IDS pada satu penderita. Mutasi tersebut menyebabkan perubahan asam amino di p.Asn265ProfsTer20. Variasi delesi satu basa nukleotida c.1023delA yang menyebabkan perubahan susunan asam amino dengan notasi p.Glu341AspfsTer19 ditemukan pada seluruh sampel. Selain itu, ditemukan pula mutasi missense novel c.1033T>C yang mengubah asam amino triptofan menjadi arginin p.Trp345Arg, dan variasi delesi satu basa nukleotida c.1041delA yang menyebabkan perubahan susunan asam amino dengan notasi p. Lys347AsnfsTer13 pada ekson 8 dari gen IDS pada satu penderita lain.
<hr>Mucopolysaccharidosis II MPS II, or Hunter syndrome, is a rare X linked recessive disease caused by lysosomal storage disorder due to the deficiency of the lysosomal enzyme iduronate 2 sulfatase IDS . The IDS gene is important for the lysosomal degradation process of dermatan sulfate and heparan sulfate, as the deficiency of the IDS gene will lead to the accumulation of these Glycosaminoglycans GAGs. Exon specific analyses on exon 6 and exon 8 of the IDS gene were done on patients with MPS II in Cipto Mangunkusumo National Referral Hospital, Jakarta, Indonesia. In this study, samples from MPS II patients and normal samples were analyzed using PCR and sequencing methods.
The results show novel mutations in Indonesian patients with MPS II. A 14 bp long insertion mutation c.792 793insCCCCTGTGGCCTAC were found on exon 6 of the IDS gene in one patient. This mutation leads to the alteration of amino acids at p.Asn265ProfsTer20. A single nucleotide deletion variant c.1023delA which leads to the alteration of amino acids in the p.Glu341AspfsTer19 was found in all patients. Other than that, a novel missense mutation c.1033T C which leads to the alteration of amino acid from tryptophan to arginine p.Trp345Arg and a single nucleotide deletion variant c.1041delA which leads to the alteration of amino acids at p.Lys347AsnfsTer13 on exon 8 of the IDS gene were found in another patient.