FOLIA MEDICA INDONESIANA

Vol. 43 No. 4 October - December 2007

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**METABOLIC SYNDROME IN PATIENTS WITH MITOCHONDRIAL DIABETES MELLITUS**

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**Abstract**

A few mtDNA mutations strongly associated with diabetes is called as Mitochondrial Diabetes Mellitus (MDM), with the causal mutation being the A3243G mutation in the mitochondrial DNA-encoded tRNA(Leu, UUR) gene. It is unfortunate that A3243G mutation was not found in Indonesia after more than a thousand of mtDNA samples being examined in Jakarta, Jogjakarta and Surabaya. In Indonesia G3316A and T3394C mitochondrial mutation was found to be the predisposing factors of DM. We investigated the contribution of Metabolic syndrome (MetS) in MDM cases related to G3316A and T3394C mtDNA mutation. Case series of three DM patients with G3316A and five DM patients with T3394C mutation were detected using Polymerase Chain Reaction (PCR) and digested with HaeIII restriction enzyme, which was screened from family study of two probands G3316A carrier and also from two probands T3394C carrier. Mutation was confirmed using DNA sequencing on the ABI 377A. Obesity was detected by Body Mass Index (BMI) based on PERKENI consensus 1998, and it is defined by BMI > 25 kg/m². MetS criteria used modified WHO 1998 criteria after adjusting BMI according to Perkeni Consensus (1998). Clinical profiles were assessed in order to show the characteristic of MDM and giving the information of complications and clinical outcomes. We found two patients with obesity and one overweight among three DM with G3316A mutations. Nutrition status showed three cases of obesity and one overweight patient from five DM with T3394C mutation. Insulin resistance was detected in two from three DM patients with G3316G mutation, and two of four T3394C mutations. All of three DM patients with G3316A mutation had low HOMA-B (23.55 ± 18.65 %) and three of four DM patients with T3394C mutation had low HOMA-B (25.91 ± 35.30 %). Hypertension was found in one of two patients with G3316A, and all of three with T3394C being examined. Dyslipidemia was shown in all of DM with mutation. Two of three patients harboring G3316A mutation showed MetS according to modified WHO 1998 criteria. The first of G3316A patient has met all MetS criteria. The second of G3316A patient had all criteria except hypertension. Three of four patients harboring T3394C mutation showed MetS according to modified WHO 1998 criteria. The first of T3394C patient has met all MetS criteria. The second of T3394C patient met all criteria except low HDL cholesterol. The third of T3394C had four positive criteria except low HDL cholesterol and microalbuminuria. One patient of G3316A mutation had cardiovascular complication and one patient of T3394C mutation had stroke known as the end result of MetS. MetS could play a role in MDM with G3316A and T3394C mtDNA mutation, which was proven by modified WHO 1998 MetS criteria among study population. MetS was found in two of three patients harboring G3316A mutation and in four of five patients with T3394C mutation. One patient of G3316A mutation has cardiovascular complication and one patient of T3394C mutation has stroke that is also known as the end result of MetS.

**Keyword**: metabolic, syndrome, mitochondrial, diabetes, mellitus, (MDM), Metabolic, syndrome, (MetS), G3316A, ,

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