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Diabetes due to the mt3243 A>G mutation among young adult diabetic subjects in Sri Lanka - prevalence and clinical heterogeneity; Abstract; Ceylon College of Physicians- 40th Annual Scientific Sessions; 2007_.64pp

Abstract : Introduction: The maternally inherited mt3243A>G mutation is associated with a variable clinical phenotype including diabetes and deafness (MIDD). The frequency of this mutation in Sri Lanka (SL) is not known. We aimed to determine the prevalence and clinical characteristics of MIDD among young adult-onset diabetes subjects in SL. Methods: DNA was available from 1007 subjects (age of diagnosis 16-40 yrs, age at recruitment </45 yrs). The cohort was screened by quantitative real time PCR on an ABI 7900HT system using sequence specific TaqMan probes. Samples with heteroplasmy >/5.0% were considered positive. Results: 9 (4 males) mutation positive subjects were identified (prevalence 0.9%). They were diagnosed at a younger age (26.0 ± 4.8 vs $31.9 \text{ yrs} \pm 5.6$, $p=0.002$) and were lean (BMI 18.6 ± 2.6 vs $25 \pm 15.0 \text{ kgm}^{-2}$, $p<0.0001$) compared to non-mutation carriers (NMCs). A combined screening criteria of any two of; maternal history of diabetes, personal history of hearing impairment and family history of hearing impairment identified only 4 (44%) of the carriers with a positive predictive value of 9%. One mutation positive subject (11.1%) had the metabolic syndrome vs 60% of NMC. Insulin therapy from the diagnosis was used in 4 (44.0%) of carriers compared to 14.3.0% of NMC ($p=0.04$). Conclusions: The prevalence of mt3243A>G mutation among young-onset diabetic subjects from SL was 0.9%. Our study demonstrates that a maternal history of diabetes and either a personal and/or family history of deafness distinguish less than half of patients with MIDD from Sri Lankan subjects with young-onset diabetes.