Ceylon College of Physicians- 40th Annual Scientific Sessions: 2007; 64p.

Diabetes due to the mt3243 A>G mutation among young adult diabetic subjects in Sri Lanka - prevalence and clinical heterogeneity

Katulanda, P; Silva, A; Mahesh, JG; Sheriff, MHR; Matthews, DR; McCarthy, MI; Gloyn, AL

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: DNAwas available from 1007 subjects (age of diagnosis 16-40 yrs, age at recruitment /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 yrs ± 5.6, p=0.002) and were lean (BMI 18.6 ± 2.6 vs 25 ± 15.0kgm-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 carries 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 youngonset 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.