ABSTRACT

It has been demonstrated that *GJB2* mutation and biotinidase deficiency are common causes for non-syndromic hearing loss (NSHL). *GJB2* gene codes for gap junction beta 2 protein (connexin 26) that helps maintain proper level of potassium ions in the inner ear which is essential for the conversion of sound waves to electrical nerve impulses Biotinidase deficiency, on the other hand, is an inherited disorder characterized by the inability of the body to reuse biotin which can cause NSHL, if untreated. Therefore this study attempts to investigate whether *GJB2* mutations and biotinidase deficiency is the major cause of NSHL in a small population of Malaysians in Sungai Petani. Blood samples from volunteers (31 with NSHL and 11 with normal hearing) were collected and subjected to genomic DNA extraction prior to PCR amplification of the Cx26 (*GJB2* gene) region. The amplicon was sequenced and analyzed for mutations. The blood samples were also subjected to biotinidase assay. Out of 42 samples, 11 had mutation which comprised of 6 from patients with NSHL while the balance 5 from normal individuals. For those with NSHL, 3 of the mutations were frameshift mutation and the rest were missense type. The most frequently observed mutation was V153I, which was found in one patient with NSHL and 4 others with normal hearing. The 35insG, 217delC, G160S and 509delA mutations were observed only in patients with NSHL with the frequency of occurrence of one patient per mutation. The most widely reported V37I mutation that is prevalent in Asian community with NSHL was also found in one of the patient with hearing loss as well as one normal individual. As for the biotinidase assay, all samples recorded normal enzyme levels. The outcome indicates that biotinidase deficiency and *GJB2* mutations were not the common cause for NSHL among the test subjects of this study, although mutations related to NSHL were observed. However, repeating this experiment using a larger sample volume would give a more comprehensive answer as to whether *GJB2* gene mutation and

biotinidase deficiency would serve as efficient marker to detect NSHL as well as understand whether it plays a role in the NSHL pathogenicity, among Malaysians.