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# Application of next generation sequencing upon the detection of 100 mutations in 18 deafness genes in Vietnamese children with non-syndromic hearing loss

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**Introduction:** There are 15000 hearing-impaired children are born in Vietnam every year. Early detection of common deafness mutations is a key factor for diagnosing, helping hearing-loss children to develop their language and awareness normally.

**Objective:** To identify the carrier rate of new common mutations and common mutations associated with congenital deafness children in Vietnam.

**Methodology:** 80 hearing-impaired and 100 normal children from Vietnam are included. Technology of next generation sequencing was applied to detect 100 mutations of 18 deafness genes, namely GJB2, GJB3, SLC26A4, MT-RNR1, MT-CO1, MT-TL1, MT-TS1, MT-TH, DSPP, GPR98, DFNA5, TMC1, MYO7A, TECTA, DIABLO, COCH, MYO15A and PRPS1.

**Result:** Identified 15 mutations of deafness gene of hearing-impaired patients (account for 18.75% in total) including: 7 cases of gene GJB2 (2 cases of homozygous mutation and 5 cases of heterozygous mutation), 3 cases of heterozygous mutation of gene SLC26A4, 2 cases of homozygous mutation of MT-RNR1, 1 case of heterozygous mutation of gene TMC1, 1 case of heterozygous of gene MT-TH and 1 case of heterozygous mutation of gene MT-TL1. No mutations were identified in the control group.

**Conclusion:** The incidence of deafness mutations in hearing-loss group is 18.75%. Mutations of GJB2 cover the largest proportion among 18 genes investigated. In this review, we describe commonly used genomic technologies as well as the application of these technologies to the genetic diagnosis of deafness and to the discovery of novel genes for non-syndromic deafness.

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