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New SNP variants of *MARVELD2* (*DFNB49*) associated with non-syndromic hearing loss in Chinese population

Key words: Chinese population, *MARVELD2*, Non-Syndromic Hearing Loss, SNP variants

Research Summary

This article conducted a case-control study in an eastern Chinese population to profile the spectrum and frequency of *MARVELD2* variants, as well as the association of *MARVELD2* gene variants with NSHL.

- **PCR+Sanger sequencing**
- **In silico Filter of Candidate Variants**
  - ✓ allele frequency (controls and ExAC)
  - ✓ amino acid conservation
  - ✓ chemical properties change of amino acid
  - ✓ detrimental prediction (SIFT, Polyphen)
  - ✓ protein domain and structure analysis (I-TASSER)

Innovation points

- **Identification** of 11 ***MARVELD2*** variants in a cohort of 283 Chinese NSHL patients. c.730G>A variant may have relatively high correlation with NSHL pathogenicity.

- **Emphasis** of the ***MARVELD2*** mutation spectrum in the Chinese population are significantly different from those reported in Pakistan, Slovak, Hungarian and Roma origin families.

Figure 1