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Clinical Phenotypic Characterization of the SLC26A4 Mutation in Pendred Syndrome/Nonsyndromic Enlarged Vestibular Aqueduct

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Abstract : Objective: The aim is to summarize the Solute Carrier Family 26 Member 4 (SLC26A4) mutations and clinical phenotypic characteristics of patients with Pendred syndrome/nonsyndromic enlarged vestibular aqueduct (PS/NSEVA). Design: A retrospective cohort study for the Chinese population was performed to analyze the hearing test results of 406 patients with PS/NSEVA who had a SLC26A4 mutation and the relationship between inner ear imaging and audiology. Results: There was a significant difference in the mean hearing threshold in patients with biallelic mutations (M2), monoallelic mutations (M1), and nonallelic mutations (M0) and between patients with isolated vestibular aqueduct enlargement (IEVA) and patients with IEVA combined with Mondini malformation. There was no significant difference between patients with different gene mutation types or different sexes or between the width of the vestibular aqueduct (VA) and the mean hearing threshold. The degree of hearing loss was linearly correlated with age. Conclusions: We propose that the presence or absence of SLC26A4 mutation, whether combined with Mondini malformation and patient age, are essential factors affecting the degree of hearing loss in the Chinese population. However, the number and type of mutations, degree of VA expansion, and sex of the patients did not affect the clinical audiological phenotype.

Keywords : hearing loss, Pendred syndrome/nonsyndromic vestibular enlargement of aqueduct, radiologic, SLC26A4
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