Novel Mutations in MYO7A and USH2A in Usher Syndrome

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Abstract:

Purpose:
Usher syndrome is an autosomal recessive disease associating retinitis pigmentosa and neurosensory deafness. Three clinical types (USH1, USH2, USH3) and 11 mutated genes or loci have been described. Mutations in MYO7A and USH2A are responsible for about 40% and 60% of Usher syndromes type 1 and 2, respectively. These genes were screened in a series of patients suffering from Usher syndrome.

Methods:
We performed SSCP screening of MYO7A in 12 unrelated patients suffering from Usher syndrome type 1 (USH1) and USH2A in 28 unrelated patients affected by Usher syndrome type 2 (USH2).

Results/conclusions:
Six mutations in MYO7A were found in five patients, including two novel mutations c.397C>G (His133Asp) and 1244-2A>G (Glu459Stop), accounting for 42% of our USH1 patients. Twelve mutations in USH2A were found in 11 patients, including four new mutations c.850delGA, c.1841-2A>G, c.3129insT, and c.3920C>G (Ser1307Stop), accounting for 39% of our USH2 patients.

Keywords:
Usher syndrome, MYO7A, USH2A, novel mutations