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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