PREVALENCE OF DELETIONS IN GJB6 GENE IN A SAMPLE OF NEONATES AND NON-SYNDROMIC BRAZILIAN DEAF PATIENTS

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Alterations in DFNB1 locus are present in 50% of families with autosomic recessive non-syndromic hearing loss. Two related genes have been cloned in this locus,the GJB2 and the GJB6 gene. Both of them encode connexins, a family of proteins involved in cochlear homeostasis maintenance. One specific GJB2 mutation, 35delG, is one of the most frequent disease mutation identified, and a problem with deaf carriers of this mutation is that in some cases only one allele is mutant, despite of the recessive pattern of inheritance. This issue was partially solved by the finding of two deletions that truncates the neighboring GJB6 gene (del-GJB6-D13S1830 and del-GJB6-D13S1854). In order to determine the deletions's frequency in brazilian population, in this study were screened both deletions and 35delG mutation in 1741 neonates. Furthermore, 711 patients with non-syndromic hearing loss were analyzed. In the sample of newborns, the deletions were not detected, what accords with other studies and proves the low prevalence of these mutations. Among deaf individuals 49 were heterozygotes for 35delG mutation, and between these we found three individuals with del-(GJB6-D13S1830) and three with del-(GJB6-D13S1854), with a prevalence of 6,12% for both deletions. After all, the deletions were relatively frequent between deaf patients heterozygotes for 35delG mutation; thus, we recommend the screening for this group. However, a newborn screening isn't necessary, due to the absence of deletions in our huge neonates population.

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