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Significance of heterozygosis M34T mutation of GJB2 gene in non-syndromic congenital deafness. Retrospective analysis of 12,472 samples of amniotic fluid.

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Abstract

OBJECTIVE: to determinate the role of heterozygosis of M34T mutation of GJB2 gene in non syndromic congenital deafness.

METHODS: retrospective study between March 2010 and June 2013. Molecular screening for 35delG and M34T mutations of the GJB2 gene was offered to all women undergoing to second trimester genetic amniocentesis. Patients were excluded from the study group if one of the following conditions were present: infections, fetal abnormalities, family history for congenital deafness, diagnosis of chromosomal abnormalities, and consanguinity between parents.

RESULTS: a total of 12.472 Caucasian women gave informed consent for this test. Seventy-seven cases were excluded. From the 12.395 amniotic fluid analysis remained, the following was found: 2 cases of 35delG homozygosis and 352 cases of heterozygous carriers (42 M34T mutation, 298 35delG mutation, 12 double heterozygosis M34T/35delG). The follow up in first year of life in the 42 newborns with heterozygosis for M34T mutation showed a mild deafness in 23 cases.

CONCLUSIONS: in our series, presence of heterozygosis M34T mutation is associated in more than 50% of cases to mild congenital deafness.

KEYWORDS: M34T mutation; amniocentesis; congenital deafness; prenatal diagnosis

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