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LETTER

On the origin and frequency of the 35delG allele in GJB2-linked deafness in Europe

In the January issue of the journal, an article entitled 'A carrier frequency of the 35delG deafness mutation in European populations'[1] was published. This mutation occurs in the connexin 26 (GJB2) gene and causes deafness in the homozygous state. The investigation covered many populations of northern, central and southern Europe, and reveals a high frequency of the 35delG allele in all populations with higher prevalence in southern Europe, and the highest percentage in Estonia. Estonian data clearly separate this population from the other investigated groups. We decided to expand this study further to the East, and investigated several populations of Russia. First, like the authors of the above article, we considered this investigation important for defining the strategy of diagnosis and genetic counselling of congenital deafness in Russia. Second, this study could clarify the origin and history of the 35delG allele. Taking into account the high frequency of this mutation in Estonia, we paid special attention to related Finno-Ugric populations, namely Mari and Komi. Three other populations, Bashkirs, Chuvashs and Yakuts, belong to Turkish speaking populations. In the ethnogenesis of Bashkirs and Chuvashs, not only was there Turkic participation but also some other populations including Finno-Ugric. This is especially true for Chuvashs. The Yakut population was the most mongoloid among the three investigated Turkic populations.

Altogether, 560 persons from five ethnic groups of Russia were analysed for the 35delG allele (see Table 1). Most of the DNA samples were used for other investigations in population genetics, and selection criteria were typical for this type of investigation. Analysis of the 35delG allele was performed in one laboratory simultaneously for all available DNA samples. We used primers 5′-CTTTTCCAGAG-CAAACCGCCC-3′ and 5′-TGCTGGTGGAGTGTTTGTTCAC-3′ for amplification of an 89 bp fragment in normal chromosomes, and an 88 bp fragment in mutated chromosomes. Normal and mutated alleles were clearly distinguished on 10% polyacrylamide gel (acrylamide:bis-acrylamide 29:1.3). The same approach is used routinely for DNA testing of patients with deafness. Twelve mutated chromosomes were found, resulting in an average carrier frequency of 1/46.7. This corresponds to the frequency of more than 1% of chromosomes with mutation.

Table 1

Ethnic group	*Number of investigated persons*	*Number of chromosomes*	*Number of chromosomes with 35delG mutation*	*Frequency (%)*
Mari	194	388	5	1.3
Komi	51	102	–	0
Chuvashs	154	308	4	1.3
Yakuts	106	212	1	0.5
Bashkirs	55	110	2	1.8

In spite of the smaller scale of the present study, it adds remarkable information to the previous investigation. The most important fact is that the 35delG allele is definitely present not only in western but in eastern Europe too, and is frequently found in Finno-Ugric and Turkic populations. The mean frequency of this mutation in the mentioned populations seems higher than in western Europe. Thus, suggestion of origination of the mutation in Europe seems doubtful. Further study of populations belonging to Ural and Altai linguistic families could shed light on the history of the 35delG mutation and will probably add some important information regarding the relationship of modern inhabitants of western Europe, Uralic and Turkic populations.

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References

1 Gasparini P *et al*: High carrier frequency of the 35delG deafness mutation in European populations. Genetic Analysis Consortium of GJB2 35delG. *Eur J Hum Genet* 2000; **8**: 19–23.