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# THE RATE AND STRUCTURE OF THE ASSORTATIVE MARRIAGES AMONG DEAF INDIVIDUALS IN THE REPUBLIC OF BURYATIA

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It is currently accepted that the reproductive capabilities of deaf people increase over time, and marriages between deaf people occur according to the principle of assortativity, which in total can lead to an increase in the frequency of one major autosomal recessive form of deafness. In this regard, the purpose of this work was to analyze the share and structure of assortative marriages based on deafness in the Republic of Buryatia. The sample of deaf individuals consisted of 201 people (113 female and 88 male) aged from 21 to 77 years (mean age 46.7 ± 7.9 years). For analysis of the marriage structure of deaf people, information was available for 168 marriages. Individuals were considered married if they had a registered marriage and/or if they had common children. An assortative marriage (AM) was a marriage in which both partners were deaf. An AM in which all the children were deaf was considered non-complementary, and an AM in which all the children were hearing was considered complementary. Marriages in which there were both hearing and deaf children were designated as segregating. An analysis of the marriage structure of deaf people showed that the proportion of marriages between deaf individuals in Buryatia is 81.8% (122 out of 149 marriages analyzed), and in 18.2% of cases (27 marriages) marriages were concluded between deaf and hearing people. Among all AM, the share of complementary marriages was 86.9% (106 out of 122), non-complementary - 5.7% (7 out of 122) and segregating 7.3% (9 out of 122). It was revealed that with a high frequency of AM, the marital structure was characterized by a high proportion of complementary and low proportion of non-complementary and segregating AM, which may be explained by the peculiarities of the genetic structure of hereditary forms of hearing loss in the Republic of Buryatia.

**Keywords:** hereditary hearing loss, assortative marriages, sign language, autosomal recessive deafness 1A (DFNB1A), Buryatia.

**Introduction.** The inventor of the telephone, Alexander Graham Bell, first suggested in 1883 that frequent marriages between deaf individuals could lead to an increase in the occurrence of hereditary forms of hearing loss. In the 2000s, this assumption was revisited by Walter Nance, who formulated the hypothesis that the reproductive opportunities of deaf individuals increase over time, and marriages between deaf individuals occur not randomly but according to the principle of assortative mating. This could lead to an increase in the frequency of the "connexin" form of deafness, caused by pathogenic variants of the *GJB2* gene. Subsequent computer modeling supported this hypothesis, showing that increased reproductive opportunities and intensive assortative mating could indeed double the frequency of the most common "connexin" deafness form in the United States in less than 200 years, after the introduction of sign language. This hypothesis was further supported by evidence from the dynamics of the socio-demographic structure of individuals with hearing impairments in the United States from the 19th to the 20th centuries, as well as from other computational experiments on the computer modeling of the spread of hereditary hearing loss.

Nance's hypothesis suggests that the effect of assortative mating is limited to the most common form of recessive hearing loss because, with frequent assortative mating, critical is the proportion of marriages in which both partners have the same genetic etiology of hearing loss and have a 100% chance of having deaf children. According to this hypothesis, such marriages are called non-complementary. Accordingly, in populations where one major recessive form of hearing loss predominates, intensive assortative mating marriages for deafness may lead to greater transmission of pathogenic alleles in subsequent generations.

Earlier studies have shown that the proportion of assortative marriages between deaf individuals in Yakutia is 77.1%, among which 24% are non-complementary (hearing loss in both spouses is caused by recessive pathogenic variants of the *GJB2* gene). Agent-oriented computer modeling of the spread of the "connexin" form of deafness in Yakutia showed that in the presence of assortative marriages, the proportion of recessive mutant homozygotes almost doubled within the first four generations. These data suggest that assortative marriages between deaf individuals in Yakutia may lead to an increase in the frequency of hereditary "connexin" hearing loss, as previously shown for the population of the United States.

Therefore, the aim of this study was to analyze the proportion and structure of assortative marriages for deafness in the previously unstudied neighboring region of Eastern Siberia - the Republic of Buryatia.

**Materials and methods.** Specialized surveys were conducted with deaf individuals belonging to the Buryat branch of the All-Russian Society of the Deaf (Ulan-Ude city) with the help of sign language interpreters. The sample of deaf individuals consisted of 201 individuals (113 women and 88 men) aged 21 to 77 years (mean age 46.7±7.9 years). The

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ethnic composition of the sample of deaf individuals was as follows: Buryats - 98 people (48.7%), Russians - 85 people (42.3%), and others (individuals of mixed ethnic backgrounds, as well as occasional representatives of other nationalities) - 18 people (8.9%).

For the analysis of marriage structure, information was available for 168 individuals, which included 149 marriages (including remarriages). Individuals were considered married if they had a registered marriage and/or shared children. An assortative marriage (AM) was considered a marriage in which both spouses were deaf. A non-complementary marriage was considered an AM in which all children were deaf, while a complementary marriage was considered one in which all children were hearing. Marriages in which there were both hearing and deaf children were designated as segregating.

Statistical analysis between groups was performed using the chi-square test with Biostatd software (McGraw-Hill, Inc. Version 3.03). Differences were considered statistically significant at $p<0.05$.

The examinations provided for within the framework of this research were conducted after obtaining informed written consent from the participants. The research was approved by the local biomedical ethics committee at the Yakut Scientific Center of Complex Medical Problems (Yakutsk city) in 2019 (Protocol No. 7 dated August 27, 2019).

**Results and discussion.** The analysis of the marriage structure of deaf individuals showed that the proportion of marriages between deaf individuals in Buryatia is 81.8% (122 out of 149 analyzed marriages), while in 18.2% of cases (27 marriages), marriages were contracted between deaf and hearing individuals. Among all assortative marriages, the proportion of non-complementary marriages was 5.7% (7 out of 122), segregating marriages 7.3% (9 out of 122), and complementary marriages 86.9% (106 out of 122) (Table 1).

We compared the proportion of assortative marriages in Buryatia with data from 10 other studied regions worldwide, for which the proportion of marriages between deaf individuals was calculated (Table 2). As a result, the proportion of marriages between deaf individuals in Buryatia (81.8%) was one of the highest, indicating the presence of positive assortative mating based on the "deafness" trait. A high proportion of assortative marriages between deaf marital partners, along with widespread use of sign language, is one of the main characteristics of consolidated deaf communities (deaf culture). A similarly high proportion of AM marriages is considered characteristic of countries where deaf people actively use sign language: Northern Ireland (UK) - 89.3%, USA - 79.5%, Sweden (Narke region) - 99%. In contrast, in countries where the introduction of sign language has occurred relatively recently, the proportion of AM is much lower: Tunisia - 10-30%, Mongolia - 37.5%, Turkey - 46.8% (Table 2).

**Table 1**

**The marital structure of hearing-impaired individuals in the Republic of Buryatia**

| Total marriages | n (%) |
|---|---|
| Всего | 149 (100) |
| Assortative marriages: | 122 (81.8) |
| - Non complementary | 7 (5.7) |
| - Segregating | 9 (7.3) |
| - Complementary | 106 (86.9) |
| Non assortative marriages | 27 (18.2) |

As seen from Table 2, two regions in Russia (Buryatia and Yakutia) also have a high proportion of AM marriages in relation to deafness, likely associated with a longer history of sign language use. For example, the first school for the education of deaf people based on sign language was esTableished in Yakutsk in 1951, whereas in Mongolia, the first similar school was opened in Ulaanbaatar only in 1995.

Next, we compared the proportion and structure of assortative marriages in Buryatia with those in Yakutia, where a

**Table 2**

**The proportion of assortative marriages in Buryatia compared to available literature data**

| Region | Proportion ofAM | References |
|---|---|---|
| Russia, Republic of Buryatia | 81.8 | [This study] |
| Russia, Republic of Sakha (Yakutia) | 77.1 | [1] |
| USA | 79.5 | [13] |
| United Kingdom, Northern Ireland | 89.3 | [17] |
| Sweden, Narke | 99.0 | [8] |
| Sweden, Varmland | 10.0 | |
| India | 56.6 | [16] |
| Turkey | 46.8 | [18] |
| Mongolia | 37.5 | [12] |
| Tunisia | 10.0-30.0 | [9] |

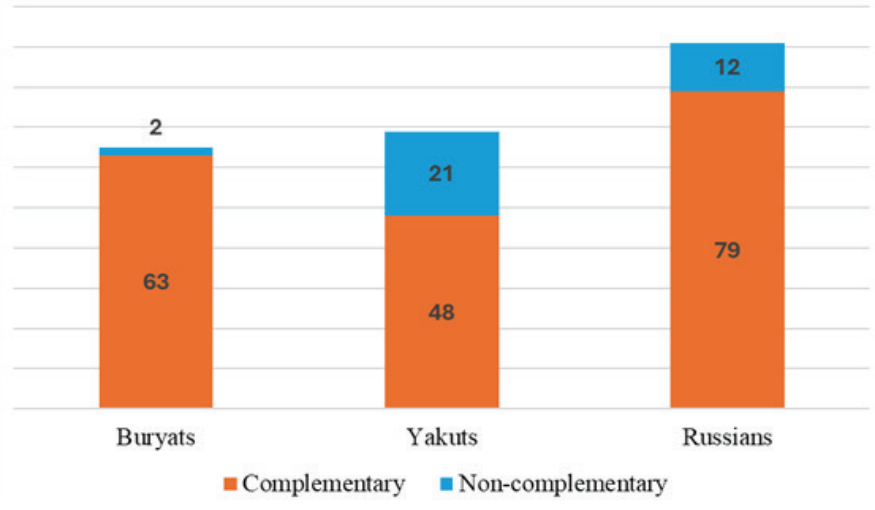
Note: Regions where the proportion of assortative marriages was below 60% are highlighted in gray.

**Table 3**

**Comparative analysis of the proportion and structure of assortative marriages for deafness in Buryatia and Yakutia**

| Region | Assortative marriages | Non-complementary marriages | Complementary marriages | Segregating | References |
|---|---|---|---|---|---|
| Republic of Buryatia | 122 out of 149 (81.8%) | 7 out of 122 (5.7%) | 106 out of 122 (86.9%) | 9 out of 122 (7.3%) | [This study] |
| Republic of Sakha (Yakutia) | 81 out of 105 (77.1%) | 19 out of 81 (23.5%) | 62 out of 81 (76.5%) | 24 out of 81 (29.6%) | [1] |
| $\chi 2$ | 0.86 | **13.69** | 3.65 | **17.71** | - |
| *p* | >0.05 | **<0.001** | >0.05 | **<0.001** | - |

Note: statistically significant differences ($p<0.05$) are highlighted in bold.




Proportion of complementary and non-complementary assortative marriages by ethnicity

Table 4

**Comparative analysis of the contribution of the connexin deafness (DFNB1A) among patients with hearing impairments among Buryats and Yakuts**

| Ethnicity | DFNB1A proportion | References |
|---|---|---|
| Buryats | 4 out of 79 (5.1%) | [19] |
| Yakuts | 29 out of 55 (52.7%) | [1] |
| $\chi 2$ | **36.69** | - |
| *p* | **<0.001** | - |

Note: Statistically significant differences are highlighted in bold (p<0.05).

previous analysis of assortative mating among people with hearing impairments was conducted [1]. The comparative analysis of the proportion and structure of assortative marriages for deafness in Buryatia and Yakutia is presented in Table 3. The proportion of assortative marriages for deafness in Buryatia - 81.8%, was comparable to the proportion of assortative marriages in Yakutia - 77.1% ($\chi 2$= 0.86, *p*>0.05). However, the structure of assortative marriages differed significantly, as the proportion of non-complementary assortative marriages in Buryatia (5.76%) was four times lower than in Yakutia, where non-complementary marriages accounted for - 23.5% ($\chi 2$= 13.69, *p*<0.001). Additionally, the proportion of segregating marriages in Buryatia (7.36%) was also four times lower than in Yakutia (29.6%) ($\chi 2$= 17.71, *p*<0.001), while the proportion of complementary marriages in Buryatia (86.9%) and Yakutia (76.5%) did not differ statistically ($\chi 2$= 3.65, *p*>0.05) (Table 3).

In the analysis of marriages by ethnic belonging, it was found that the maximum proportion of non-complementary assortative marriages was identified among the Yakuts (30%, 21 out 69 AM), while average values were found among the Russians (13%, 12 out 91 AM), and the minimum proportion of non-complementary assortative marriages was identified among the Buryats (3% - 2 out 65 AM) (Figure).

The significant difference in the proportion of non-complementary and segregating marriages, despite the similar frequency of assortative marriages for deafness in Buryatia (81.8%) and Yakutia (77.1%), may be explained by the peculiarities of the genetic structure of hereditary forms of hearing loss in these neighboring regions of Eastern Siberia. When comparing the contribution of pathogenic variants of the *GJB2* gene, which underlie the "connexin" form of hearing loss (DFNB1A, OMIM #220290), it was found that among Buryat patients (5.1%), its contribution was ten times lower than the contribution of this form of hearing loss among Yakut patients (52.7%) ($\chi 2$=36.69, *p*<0.001) (Table 4).

Previously, it was shown that the relatively high proportion of non-complementary and segregating marriages in Yakutia is associated with the prevalence of a major "connexin" form of hearing loss in this region of the world (47% of non-complementary marriages were registered among deaf individuals with DFNB1A) [1]. Currently, this region of Eastern Siberia is known as the largest local hotspot for the accumulation of the allelic form of this disease, associated with the pathogenic variant of the c.-23+1G>A splicing site of the *GJB2* gene, which formed over 800 years ago as a founder effect [6]. Analogous to the previously obtained results in Yakutia, the low proportion of non-complementary and segregating assortative marriages in Buryatia likely suggests the absence of dominance not only of the "connexin" form (contribution of DFNB1A - 5.1%, one of the lowest contributions in the world) [19], but also of any other predominant autosomal recessive form of hearing loss in the Buryat population. In turn, this almost completely negates the effect described by Walter Nance of fixation of pathogenic autosomal recessive alleles associated with assortative marriages for deafness among the indigenous population of the Baikal region.

**Conclusion.** In the Republic of Buryatia, a high proportion of assortative marriages (AM) for deafness (81.8%) was identified, indicating a high consolidation of deaf communities. Despite the high frequency of AM, the marriage structure was characterized by a high proportion of complementary (86.8%) and a low proportion of non-complementary (5.7%) and segregating assortative marriages (7.3%), which is likely associated with the peculiarities of the genetic structure of hereditary forms of hearing loss in the Republic of Buryatia.

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# AGE AND GENDER CHARACTERISTICS OF THE CHEMICAL ELEMENTS IN THE HAIR OF RESIDENTS OF UFA CITY

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The formation of the elemental status of the human body living in a certain region de-pends on the biogeochemical, ecological and geographical factors of the area, nutrition, and pro-fession. The content of chemical elements in the body and their ratio are an indicator of human health. For residents of the city of Ufa, a large industrial center of the Republic of Bashkortostan, References intervals for the concentration of macro- and microelements in hair have been developed. However, to identify deficiencies, excesses or imbalances of chemical elements in the body, it is necessary to take into account gender and age differences. In this regard, the purpose of this work was to study the age and gender characteristics of the content of macro- and microelements in the hair of residents of a large industrial city. The content of 12 chemical elements in the hair of residents of the city of Ufa of the age groups - 18-29 years, 30-44 years, 45-65 years and over 65 years old was determined by atomic absorption spectrometry using devices with flame and electrothermal atomization.

In all age groups, a deficiency of zinc and copper was detected in the hair of both men and women against the background of excess chromium, lead and manganese. Median concen-trations of iron, magnesium, cadmium, mercury and arsenic are within the 25-75 centile range. A deficiency of calcium in the hair of men under 65 years of age was revealed; in women it was found in the range of References values. An excess of 4-11% of the upper limit of permissible Ni content in men was esTableished. There is a tendency towards higher accumulation of cadmium, lead and arsenic in the hair of the male population. In women over 65 years of age, the minimum levels of essential elements are determined - calcium, magnesium, zinc and copper, in men of this age - copper, iron and manganese. Toxic elements cadmium, lead and arsenic accumulate to their maximum at 65 years of age and older.

The results of the study can be used as an additional method for determining the charac-teristics and time of onset of various diseases, as well as for the medical correction of diselementoses and enriching diets with essential micronutrients.

**Keywords:** macroelements, microelements, hair, age, residents of Ufa.