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THE ANALYSIS OF SOCIAL READINESS OF PREGNANT WOMEN FROM FAMILIES BURDENED WITH HEREDITARY SPINOCEREBELLAR ATAXIA TYPE 1 TO UNDERGO PRENATAL DNA TEST

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ABSTRACT

The analysis of social readiness of pregnant women from burdened families to undergothe prenatal DNA diagnostics program of spinocerebellar ataxia type 1 (SCA1) neurodegenerative monogenic disease is provided in the article. The research opens some social problems of pregnant women with risk of SCA1 for generation that will allow defining further competent tactics of the medicogenetic and social help to the families burdened with SCA1.

Keywords: prenatal diagnostics of hereditary diseases, spinocerebellar ataxia type 1, social readiness, tainted families.

Introduction

Spinocerebellar Ataxia Type 1 (SCA1) – an incurable, hereditary, autosomal dominant monogenic disease of the motor system, which is mainly late-onset and the general symptom is dystaxia resulting from cerebellum lesions, its communications and the corresponding sensory systems [1,3].

Frequency of SCA1in Yakutia is 46 cases on 100 thousand people of rural population in comparison with 1-2:100 thousand people of the world population. In connection with SCA1 high prevalence in the Yakut population and with the lack of effective treatment, great attention is paid to primary prenatal wellness [5, 7].

Since the beginning of the 21st century, in practical health care of Yakutia there was a possibility of prenatal DNA testing of neuromuscular, hereditary degenerative diseases that gives the chance to future parents to make important decisions, concerning pregnancy prolongation. With the implementation of modern medical technologies and the programs of hereditary diseases prenatal screening in applied medicine of the Republic of Sakha (Yakutia), there are ethical and social issues [2, 4, 7].

The **purpose** of the research is the analysis of social readiness of pregnant women of the familiestainted with SCA1 to take part in prenatal DNA diagnostics of one of the most widespread neurodegenerative diseases in Yakutia – SCA1, so that will allow to define further competent tactics of medicogenetic and social help.

Materials and methods of the research

According to the register of hereditary and congenital diseases of the medicogenetic center №1 of the National Center of Medicine of the Republic of Sakha (Yakutia), 1197 patients, from whom 252 people with Spinocerebellar Ataxia Type 1, were under regular medical check-up in the group of hereditary diseases with the autosomal dominant mode of inheritance [8].

The analysis of personal information of 77 pregnant women who addressed on prenatal medicogenetic consultation from the burdened families with SCA1 is carried out.

Sociological (documentary, biographical) analysis methods and Sturges' statistical methods were used in the research. Data of the social relations of pregnant women were collected by direct interviews. Registration card poll specially developed for this purpose and data logging obtained from respondentswere held, according to the parameters and demands to reliability while using this method.

Results and discussion

In the research there were investigated 77 pregnant women from the burdened families with SCA1 living in the territory of the Republic of Sakha (Yakutia) aged from 19 up to 42 years. Average age of the interviewed women was 27,1±6,06 years, and 5,2% of women were 35 plus.

Along ethnic lines, 89,6% women are Sakha, 6,5% - Evenks, 3,9% - admixed nationality (half-breed). In connection with high probability of mixture of half-breed, this indicatorwas not taken into account in the final analysis of data.

It should be said,that the respondent women have rather high education level: 53,2% with higher education, 11,7% - non-complete higher education, 35,1% - advanced education. Among women with the higher education experts of social orientation – doctors and teachers prevail.

Based on the information aboutmarital status, it is possible to speak about a positive situation of the respondents: it is a first marriage of 4/5 women and this indicator positively affects the general vital activity of a person.

Surveying a **financial positionof the respondents** it should be noted, that the respondents assess their financial position as an average (72%).Average monthly earnings of the examined families varies from 40 to 50 thousand rubles. The main sources of income are the salary from the main work, casual earnings, self-employment, child allowance, and other types of grants.

From among 77 pregnant women, there are 51 (66,2%) of them agreed to undergo a prenatal test (**Group A**), 26 (33,8%) pregnant women refused conducting prenatal testing (**Group B**).

From the total number of the participants in the research, 50 (64, 9%) women are from the families tainted with SCA1 (**Group 1**), and 27 pregnant women (35,1%) are spouses of the men from the tainted families (**Group 2**).

Following the results of pre-symptomatic DNA testing, five men (18, 5%) of **Group 2** are healthy and weren't carriers of a mutant gene, so that they didn't participate in further researches.

Analysis of pregnant women who underwent prenatal DNA testing. Among 46 pregnant women who underwent prenatal DNA testing – 31 women (67, 4%) are presymptomatic bearers of a mutant gene (**Group A1**), and 15 (32,6%) of them have husbands from the tainted families (**Group A2**).

22 pregnant women have negativeresult of prenatal DNA testing (47, 8%); in all cases there was prolongation of pregnancy. There are 14 women with

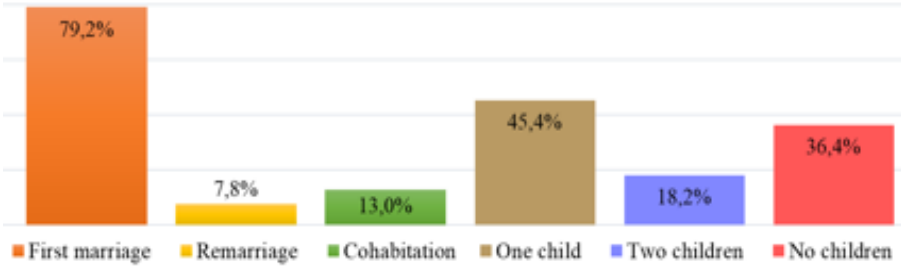

Fig.1. Marital status and parental status.

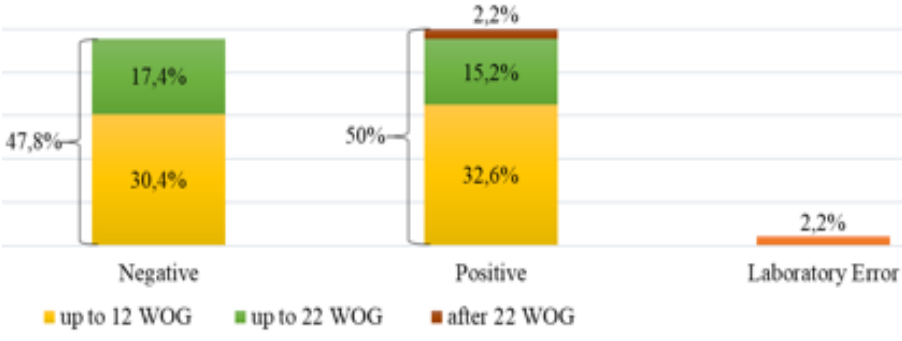

Fig. 2. DNA test results in relation with gestational age.

gestational age up to 12 weeks (63, 6%), up to 22 weeks of pregnancy – 8 women (36, 4%) (Fig. 2)

23 pregnant women have positiveresult of prenatal DNA testing (50%), from whom five families (21, 7%) made the decision on pregnancy prolongation, and in 18 cases (78, 3%) the decision on abortion was made. Durations of gestation: 15 women were at 12 weeks of pregnancy (32, 6%), 7 women – at 22 weeks (15, 2), and one pregnant woman – over 22 weeks (2,2%).

In one case (2, 2%) the analysis prenatal DNA didn't carry out because of a laboratory error (Fig. 2).

Figure 2. DNA test results in relation with gestational age

26 pregnant women refused from conducting invasive testing, from whom 19 (73, 1%) are presymptomatic carriers of a gene of SCA (**Group B1**), and 7(26,9%) women's husbands are a carrier (**Group B2**).

In our opinion, getting of presymptomatic test by a woman during the current pregnancy 12 (46, 2%) (**Group B1**) was a cause of refusals in most cases. 9 9 women (34, 6%) (**Group B1, B2**) had a strong reluctance to know the result of prenatal testing; absence on the procedure without reason explanation – in 5 cases (19,2%) (**Group B1, B2**).

Conclusions

The conducted researches taped that most of the respondents (51 person; 66,2%) are ready to undergo prenatal DNA test.

It is found that the education level practically does not influence making decision on getting prenatal DNA test. The respondents with the higher education in Group A were 58, 7%, and in Group B – 77%.

In **Group A** there are registered marriages in 36 families (78, 3%), and cohabitation or repeated marriages (unstable marriages) – in 10 families (21, 7%). In **Group B** there are registered marriages in 20 families (76, 9%), and repeated or unstable marriages, cohabitation – in 6 families (23, 1%).

By the results of the research, social readiness to participate in prenatal DNA testing in the families where women are carriers of a gene (**Group 1**) prevails over prenatal appealability in families, where husbands is representatives of families with SCA1 (**Group 2**).

The major factor of social readiness defining prenatal behavior in families with SCA1 is the traditional and historically developed relation to a disease in the tainted families. Women from **Group A1** are more motivated for prenatal DNA testing. In **group B2** refusal factors are uninformedness, family's lack of knowledge about the hereditary nature of the disease, terrify of social stigma.

The informedness events of the tainted families for the female line (**Group A1**), which were held earlier, have a positive impact on the activity of women and social readiness to undergo prenatal DNA testing.

In this regard, there is a need of development of the program for informing and rising of appealability of men (**Group 2**) who are representatives from the families tainted with SCA1 to prenatal medicogenetic consultation.

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THE PROBLEM OF SKILLS FORMING ON REPRODUCTIVE HEALTH OF GIRLS IN THE SAKHA REPUBLIC (YAKUTIA)

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ABSTRACT

The authors report a survey of respondents on reproductive health of girls. Retrospective study includes 176 respondents. According to the results of the study, it was revealed that young people had insufficient knowledge of contraception, as well as psychological readiness for the artificial termination of pregnancy. The study identified the main factors in reducing the reproductive potential of girls of fertile age.

Keywords: reproductive health, girls, contraception, abortion, sex education, the Arctic, Yakutia.

Introduction

The Republican authorities set the goal to reach one million people by the centenary of the Yakut ASSR formation (resolution of the government of the Sakha Republic (Yakutia) of December 25, 2013 No. 461). According to the Federal service of state statistics, the number of permanent population of the Sakha Republic (Yakutia) for 2017 amounted to 962.8 thousand people - these data show us that Yakutia has not retained its million status [1]. According to statistics for pre vious years - the population reached the number of 955.9 thousand people in 2012. Despite the active introduction and implementation of measures and programs to improve the birth rate in the country and particular in the Sakha Republic (Yakutia), there is no expected population growth. The children born in the 1990s during the demographic crisis have entered reproductive age, which also aggravates the demographic situation in the Sakha Republic at the moment. Nevertheless, a million Yakutians by 2022 is considered a very real goal. One of the ways to achieve this goal we see in the implementation of the preserving principle of the reproductive health of young people, especially young girls, starting from adolescence [2].

Materials and methods of research. We conducted a survey of the somatic and reproductive health of girls. A total of 176 girls aged 17 to 22 of different ethnic backgrounds were interviewed (table 1). The survey was conducted on the basis of higher educational institutions in Yakutsk. The questionnaire contained: social and anamnestic data. When collecting anamnesis, attention was paid to: the nature of menstrual function, the characteristics of sexual life, contraception, transferred gynecological diseases, previous surgical interventions, reproductive function. Statistical analysis of the study results was carried out using the programs: "Offce Microsoft Excel 2010".

Results of the study

According to the state of somatic and reproductive health of the examined girls: the leading positions in the structure of somatic diseases are diseases of the respiratory and gastrointestinal tract - every third or fourth. The Evenk girls often suffered from cardiovascular diseases compared to Yakutian and Russian girls.

Assessment of menstrual function showed that the age of menarche ranged from 12 to 14.5 years. The duration of menstruation varied from 3 to 6 days, averaging 4.1±1.5 days. In the structure of menstrual dysfunction in girls of all groups, dysmenorrhea predominated. Each eleventh girl has signs of polymenoray. Oligomenorrhea among Russian girls - every fourth, Yakutian girls - every fifth, which is significantly less when compared with Evenk girls.

Evaluation of the gynecological diseases structure has shown that the leading position is occupied by benign diseases of the cervix and inflammatory diseases of the uterus and appendages.

In the questionnaire data it is indicated that every second mother smokes in the girls'families (51.2%), 15.1% of respondents admitted in tobacco smoking, in the Yakut and Evenk group (31.7%) there is a tendency to increase the number of smokers compared to Russian (24%) and Evenks (27%). Among the reasons that prompted to start smoking: 80% of respondents indicated

Table 1

Respondents

Nationality	Amount	Part, %	Average
the Russians	21	11,9	19±1,3
the Yakuts	128	72,7	20±1,4
the Evenks	19	10,8	20±1,0
others	8	4,5	19±2,2