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MEDICAL AND SOCIAL CHARACTERISTICS OF PATIENTS WITH SPINOCEREBELLAR ATAXIA TYPE 1 WHO RECEIVED SUPPORTIVE CARE WITHIN THE CONFINES OF THE CNDD OF THE YSC CMP

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A retrospective analysis of the data of patients with spinocerebellar ataxia type 1 who received supportive care at the Center for Neurodegenerative Diseases at the YSC CMP Hospital for the period from September 2017 to June 2019 was performed. Data on age, educational and marital status are presented, and in particular, an analysis of the change in type of professional activity and family status associated with the period of pronounced manifestation of the SCAI clinic was carried out. During the period described, 52 patients received supportive care, which confirms the demand for supportive care among patients with SCAI. Low referral of patients from SCA1 endemic regions is associated with financial and climatic transportation difficulties of affected families and low awareness of specialists on-site. Of 6 patients under 35 years of age, 5 patients had inherited the mutant gene on the paternal side, which predetermined the early onset of the disease. Of the 8 patients over 60 years of age, seven had inherited the disease through the maternal line, which explains the later onset of manifestations and the more benign course of SCAI. A woman's awareness of the presence of a mutant gene for SCAI does not change her plans for childbirth. Perhaps the disease indirectly affects the change in the marital status of patients, in particular the number of divorces in women. In order to slow down the progression of the disease, ensure medical and social rehabilitation, improve the quality of life and social status of patients with SCAI, it is necessary to organize the provision of regular supportive care, monitor timely presentation at MSE, and provide full medical, social and psychological assistance on-site.

Keywords: Neurodegenerative disease, spinocerebellar ataxia, supportive care, social characteristics.

Introduction. Hereditary spinocerebellar ataxias are heterogeneous neurodegenerative diseases, the main characteristic of which is impaired coordination of movement due to degeneration of the cerebellum, afferent and efferent neuronal systems. To date, according to the transmission mechanism in generations, hereditary ataxia is divided into autosomal dominant, autosomal recessive, X-linked forms and ataxia with a mitochondrial type of inheritance. And also sporadic forms with an unidentified or unknown mechanism of genetic transmission are distinguished [1].

One of the most common hereditary neurodegenerative diseases in Yakutia is spinocerebellar ataxia type I (SCAI) (4). The frequency of SCAI in Yakutia is 46 cases per 100 thousand of the rural population, compared to 1-2:100 thousand in the world population [2,4]. The absence of a state support program for patients with hereditary ataxia in particular, and neurodegenerative diseases in general, predominantly late manifestation, severe progredient course, incurability, shortage of medical and nursing staff, and an ever increasing load due to urgent patients in specialized neurological departments, generate a lot of medical, epidemiological, organizational, ethical, social and psychological issues.

In order to implement the Decree of the President of the Russian Federation No. 204 "On National Goals and Strategic Tasks of the Development of the Russian Federation for the period until 2024" dated 07.05.2018 - the Center for Neurodegenerative Diseases (CNDD) with the NDD laboratory was created by order of the Director of the YSC CMP No. 08-06/295A at the Federal State Budget Scientific Institution YSC CMP from November 1, 2018.

An order of the Minister of Health of the Republic of Sakha (Yakutia) No. 01-07/184 dated 14.02.2019 “On the routing procedure for neurological patients suffering from neurodegenerative diseases at the outpatient and hospital stages” was also issued in order to improve the provision of specialized neurological care for patients with neurodegenerative diseases, residents of the Republic of Sakha (Yakutia) at the outpatient and hospital stages, based on the Decree of the Head of the Republic of Sakha (Yakutia) dated 27.12.2016 No. 1637 “On approval of Statutes on the Ministry of Health of the Republic of Sakha (Yakutia) and its board, and pursuant to the order of the Ministry of Health of the Russian Federation on 15.11.2011 No. 926n “On approval of the Process of provision of medical care to adults with diseases of the nervous system” [5]

With the opening of the Center for Neurodegenerative Diseases within the confines of the YSC CMP Hospital, systematic supportive care for patients with SCAI became more accessible, which made it possible to provide better medical care and draw attention to the many diverse problems of patients with hereditary ataxia in Yakutia.

Aim of the study: medical and social analysis of patients and development of

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principles for the provision of supportive care to patients with SCA1.

Materials and methods. The study was conducted at the YSC CMP Hospital in the Department of Neurodegenerative Diseases. For retrospective analysis, medical histories, genetic maps of patients with SCAI, their survey, medical and genetic counseling results and analysis of the pedigree of patients who were on regular supportive care (SC) were used. Predominantly all patients were from known pedigrees, and most of them were previously tested for the presence of the SCA1 mutation in the MGC of the RH No. 1 - NMC. The work also used statistical and analytical research methods.

Results and discussions. During the period described, 52 patients received supportive care: 18 were men (34.6%) and 34 - women (65.4%). Out of 52 patients, 17 people (32.7%) lived in the city of Yakutsk, and 35 (67.3%) in other regions of the Republic of Sakha (Yakutia). Before the official opening of the Center for Neurodegenerative Diseases (CNDD), from September 2017 to October 2018 (14 months), 31 patients received supportive care, and for the six-month period after the opening of the CNDD, there were 51 patients, which averaged 2.2 hospitalizations one month before , and 7.3 hospitalizations of patients with SCAI after the opening of the Center of NDD. This fact confirms the demand for supportive care among patients with SCAI.

Referral of patients to SC from Yakutsk hospitals was as follows: 6 patients were referred from hospital No. 3, 5 patients from the Medical Center of Yakutsk, 4 patients from hospital No. 1, and one patient from the YSC CMP Hospital. Hospitalization of patients from regions of the Republic of Sakha (Yakutia) is presented in Fig. 1. Low referral of patients from SCA1 endemic regions is associated with financial and climatic transportation difficulties of affected families and low awareness of specialists in district hospitals, outpatient clinics and paramedic stations in the villages.

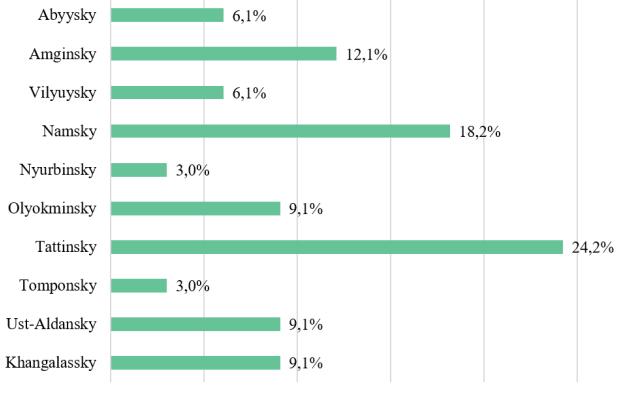

Fig. 1. Structure of referral of patients on supportive care across the Republic of Sakha (Yakutia)

Considering the late-manifesting factor of spinocerebellar ataxia type 1, an analysis of the age of the subjects is of particular interest (Fig. 2). Of 6 patients under 35 years of age, 5 patients had inherited the mutant gene on the paternal side, which predetermined the early onset of the disease. Of the 8 patients over 60 years of age, seven had inherited the disease through the maternal line, which explains the later onset of manifestations and the more benign course of SCAI.

An analysis of the educational level of patients who underwent SC showed that 27 participants in the study had higher education, which amounted to 51.9%, with secondary vocational education - 22 (42.3%) and with a secondary general education - 3 patients (5.8%). 31 (63.2%) out of 49 patients were working in a specialty relevant to their education. Since the onset of severe manifestations of the disease, 35 (68%) patients switched to light work. Seven (13%) patients had to leave work since the onset of severe manifestations of the disease. Among patients there were two researchers with a scientific degree, two physicians. Three of the patients worked in the municipal government before the onset of severe manifestations of SCAI.

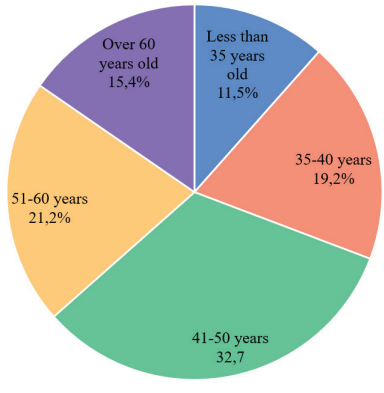

Fig. 2. Age structure of patients that have received supportive care

An analysis of marital status revealed that of the total number of divorcees, only two marriages broke up before the onset of the manifestation of the disease (Fig. 3). It is worth noting that men are more likely than women to get divorced after the onset of the manifestation of the disease in his wife. Thus, the number of divorces among male patients is 1.5 times less than among women with SCAI. The presence of children in patients was distributed as follows: did not have children - 6 patients (11.5%), one child - 10 families (19.2%), two children - 15 patients (28.8%), 3 children - 14 families (26.9%), 4 children - 4 families (7.8%) and five children were in three families (5.8%). It should be noted that 15 women from 21 families with three or more children were carriers of the mutant gene, which confirms the conclusion about extended reproduction in families with SCA1 (4).

The majority of patients who received SC repeatedly underwent the MSE examination procedure to obtain a disability class based on their spinocerebellar ataxia type I. Thus, 14 patients had a class I disability, which accounted for 26.9%, 21 patients - class II (40.4%), class III - 13 (25%) and at the time of hospitalization on SC - 4 patients had no disability class (7.7%). The results of the analysis showed that among patients with a class I disability there were 11 women (32.3% among women), and only 3 men (16.7% among men). This fact is probably due to the persistence of the female half of patients in obtaining disability classes.

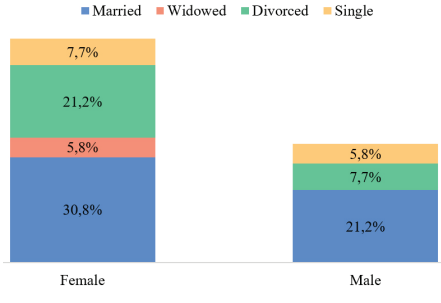

Fig. 3. Marital status of patients that have received supportive care

Conclusion. A more than a threefold increase in the frequency of hospitalizations for supportive care in the short period of time since the opening of the Center of NDD confirms the demand for SC in patients with SCAI. The analysis showed that most patients with SCA1 have higher education. A woman's awareness of the presence of a mutant gene for SCAI does not change her plans for childbirth. Perhaps the disease indirectly affects the change in the marital status of patients,

in particular the number of divorces in women. Women were more active and persistent in obtaining disability classes. In order to slow down the progression of the disease, ensure medical and social rehabilitation, improve the quality of life and social status of patients with SCAI, it is necessary to organize the provision of regular supportive care, monitor timely presentation at MSE, and provide full medical, social and psychological assistance on-site.

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