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INDICATORS OF QUALITY OF LIFE AND COMMUNICATION OF PATIENTS WITH TYPE I SPINOCEREBELLAR ATAXIA

ABSTRACT

The quality of life (QOL) and the communication ability of 41 patients with DNA verified cases of spinocerebellar ataxia type I (SCA1) was assessed with the help of the SF-36 Questionnaire. The physical, psycho - emotional parameters of QOL and the communication ability of the patients with SCA1 with different frequencies of CAG repeats were revealed, and the relationship between the revealed parameter and frequency of CAG repetitions was clarified. The cause-effect relationships of the established QOL data of the patients with SCA1 are discussed with the level of mutation in the SCA1 gene and the need to take into account the revealed features of psycho-emotional state of the patients with SCA1 in treatment and care at home.

Keywords: spinocerebellar ataxia type I, frequency of CAG repeats, mutation in the SCA1 gene, quality of life, physical and role functioning, emotions, physical and mental health, disability, social functioning, communication ability.

Spinocerebellar ataxia type I (SCA1), an autosomal dominant lesion of the cortex, conducting pathways, cerebellar and trunk nuclei, and other parts of the brain is rare in Russia (0.1-0.4 per 100,000) [13,14,16]. The development of the disease is caused by the expansion of tandem trinucleotide CAG repeats and is associated with a mutation in the SCA1 gene, on the short arm of the 6th chromosome pair, which codes the synthesis of an abnormal ataxin protein [5,11]. The onset of the disease depends on the rate of accumulation of this protein in the cells of the cerebellar cortex, which shows up with instability or awkwardness while fast walking or running after 30 years of age border. Gradually, the gait becomes "like a drunk's one," clear signs of impaired coordination of movement like changes in handwriting, speech (dysarthria), swallowing develop and tendon reflexes increase, pathological foot and carpal signs and clones, spasticity appear. A malignant form is characterized by early onset (under 20 years of age) and rapid progression. The late stage can lead to aphagia, aphonia, decreased sensitivity, loss of reflexes. In the final, patients with SCA1 are not able to walk, serve themselves and can suffer from atrophy of optic nerves, cachexia and pelvic disorders. Until the end of life, patients still retain intelligence. Yakutia is the largest source of accumulation of SCA1 (38 per 100 thousand) [1]. It should be noted that despite the above-mentioned high level of study of the causes of the neurological picture of the disease, there is no integral evaluation of the physical and psychological state of patients with SCA1, carried out carefully taking into account the frequency of their CAG repeats so far. In this regard, it should be noted that in the United States and Europe the Sort Form Medical Outcomes Study or SF-36 Questionnaire is used to assess the quality of life (QOL) parameters for this purpose. SF-36 Questionnaire with

Instruction for Conducting Interviews (Evidence - Clinical and Pharmacological Studies) and formulas for calculating QOL parameters (<http://www.sf-36.org/nbscalc/index.shtml>) [4,10] have been translated into Russian and approved by Institute of Clinical and Pharmacological Research in Russia. This allows us to use it to assess QOL of patients with chronic diseases [2, 3, 6 -12, 17].

The objective of the study was to collect data on QOL of patients with a DNA verified diagnosis of SCA1 treated by ISRH, NEFU specialists and the analysis of the data. The purpose of the study was the integrated assessment of QOL of patients with SCA1 used in the work along with an assessment of their physical, psycho-emotional abilities at different frequencies of CAG repeats, which allowed to reveal the relationship between the onset of development of the main manifestations of the disease and the CAG repeats, which would allow to objectify the causes of emerging manifestations of the disease and changes in QOL parameters of SCA1. This approach to the study of patients with SCA1 is an urgent scientific task, the implementation of which is of practical interest for specialists engaged in developing measures to optimize the treatment of patients with SCA1 in the Yakutia region being a location of accumulation of this pathology.

MATERIALS AND METHODS OF RESEARCH

The questioning of the SCA1 patients of ISRH, NEFU was carried out on the standard questions of 8 sections of the SF-36 Questionnaire according to its Instruction [4] in the conditions of leaving to the communities where the patients came from. This article describes the parameters of QOL of 41 patients with SCA1 who agreed to participate in the studies using the SF-36 Questionnaire as a group with a DNA verified SCA1 (the frequency of CAG repeats ranged from 41 to 53). The

questionings were conducted after another neurologic examination and analysis of the clinical course of the disease according to the data stored in the ISRH, NEFU archive and case records of the respondents. Five patients with severe speech disorders, who were unable to independently answer the SF-36 questions, were interviewed with the help of close relatives caring for the patients. According to the methodology of Oslo, the respondents were able to clarify the ability to establish social communications by the number of people close to them and their ability to attract the attention of people from outside, as well as to be helped by their neighbors. We analyzed the collected data taking into account the characteristics of the neurological symptomatology and the anamnesis of the disease of each patient. As it had been previously known, those patients suffering from SCA1 came from small communities (with a population from 300 to less than 400 people) due to the peculiarities of the lands that are unable to be reached by any means of transport and located far from the main roads and medical centers. In one of such communities with 350 residents as a control group 47 people whose family members had not been registered for hereditary neurologic disease according to the information of the neurological service of the district and the data of the district hospital that presented medical assistance to the inhabitants of this community were questioned. All the respondents in the control group agreed to participate in the surveys. Their age ranged from 18 to 68 years. All the data obtained in the studies were statistically processed using SPSS 22packages.

RESULTS AND DISCUSSION

The frequency of CAG repeats among the interviewed SCA1 patients was 46.89 ± 1.12 (the norm is under 35-36) and the age of the SCA1 patients varied from 18 to 69 years and was on average 46.67 ± 2.05 years. Analysis of QOL of the patients

on scales showed that the Physical Functioning (PF) scale, reflecting the degree to which the physical conditions of the SCA1 patients is limited, the physical exercise point (self-care, walking, climbing stairs, carrying heavy loads, etc.), calculated according to the Instruction of the SF-36 Questionnaire [5], was 25.38 ± 4.34 . So among the SCA1 patients the PF index was 1/4 of that of the control group, equal to 98.78 points, and thus, the PF of the SCA1 patients was sharply low compared to the data (500) in Yakutsk residents (79.7) [7], who were not suffering from this pathology. In this case the PF of the SCA1 patients was inversely dependent on the frequencies of their CAG repeats ($r = -0.612$ with $P = 0.001$ by Pearson) and the age at which dysphagia developed ($r = -0.42$ with $P = 0.05$ by Pearson) and disability ($R = -0.352$). These data proved that the ability of the physical functioning of patients, the onset of the development of swallowing disorders (dysphagia) as well as the overall age of their disability on the whole are dependent from the level of mutation in SCA1.

The value of Role-Physical Functioning (RP) of QOL of the SCA1 patients, reflecting their ability to perform usual duties, conditioned by physical condition (habitual work, performance of daily duties) was 15.28 ± 4.20 points. The revealed RP of the SCA1 patients was 1/6 of that scale in the control group (95.73 points), illustrating a 6-fold reduced level characteristic for the control group and 4.81 times less data (500) in the Yakutsk residents who were not SCA1 patients (73.6 points [7]). The analysis of the correlation of RP of the SCA1 patients showed a negative relationship with CAG repeats frequencies ($r = -0.485$ at $P = 0.05$ by Pearson). Thus, it was found that the higher the frequency of CAG repeats among the SCA1 patients, the lower their ability to perform usual duties. The presence of the direct, highly reliable relationship between the RP scale ($r = +0.632$ at $P = 0.001$ according to Pearson) with the levels of their PF scales reflected the close connection of the indices of physical abilities of the SCA1 patients. The RP scale level of the SCA1 patients appeared to be directly connected with their abilities to get social assistance, i.e. to draw the attention of people from outside and to get help from neighbors (at $r = +0.480$ with confidence $P = 0.001$ by Pearson).

The Pain Intensity scale (Bodily Pain - BP) and its impact on the ability to be occupied by daily activities, including domestic and out-of-home work, showing a limitation of pain in the ability of the SCA1 patients to be active was 70.42 ± 5.43 points. The revealed data showed that the intensity of pain in the SCA1 patients did not significantly limit their ability

to do housework and beyond. Although these data differed little from the data of the control group (76.25 points), as well as the data of the urban population (Yakutsk residents 78.00 points [7]), but a detailed analysis of the pain data of the SCA1 patients showed that the proportion of the SCA1 patients suffering from pain was 1.73 times more (40.54%) than among the control group (23.4%).

The value of QOL of the SCA1 patients on the scale of General Health (GH) was 1.5 times lower (equal to 40.41 ± 2.08 points), than that (61.76 points) in the control rural and urban population groups data 61.3 points [7]). As a result, the assessment of the condition among the SCA1 patients on the scale of the General Physical Health GH was equal to 65.43% of the control group index.

The Vitality scale -VT showing the feeling of full strength and energy or exhaustion of the SCA1 patients was 28.85 ± 4.50 points and indicated the presence of complaints, typical of lassitude, energy loss and weakness. The VT index in the control group was 91.67 points, and in the city residents group - 62.9 points [7]. So we established that the level of vitality -VT of the SCA1 patients was more than 3.17 times lower than in the control group, and in comparison with the urban residents 2.18 times lower. At the same time, the VT of the SCA1 group was highly correlated with the scales of their physical ($r = +0.685$ for Pearson $P = 0.001$) and role functioning RP ($r = +0.647$ at $P = 0.001$ by Pearson). Thus, there is a direct connection between VT and PF and RP of the SCA1 patients. Also there were direct connections between the VT scale of the SCA1 patients and the number of their close relatives ($r = +0.450$ Pearson $P = 0.001$) and assisting people ($r = +0.382$ Pearson $P = 0.05$) and neighbors looking after them ($r = +0.496$ Pearson $P = 0.001$). There was a direct relationship between the VT level and the scale of social functioning (communication) of the SCA1 patients (with $r = +0.667$ at $P = 0.001$ according to Pearson) (see below). So, the level of VT with SCA1 directly depends not only on physical, but affects social communications (see below).

Although the scale of Social Functioning (SF), showing the level of communication of the SCA1 patients, was 49.01 ± 6.32 points and 1.5 times less than the data of the city residents (74.7) and amounted to 1/2 of such control (98.69 points) but with high reliability (according to Pearson $P = 0.001$) was associated both with PF ($r = +0.681$), RP ($r = +0.647$) and with VT as it was indicated. The frequency of CAG repeats in the SCA1 patients had a negative effect on the onset age ("debut") of the disease ($r = -0.673$ at $P = 0.001$) and the onset of speech impairment ($r = -0.680$ at

$P = 0.05$), almost as much as on the age of the beginning of the coordination disorder (unsteadiness of gait $r = -0.600$, awkwardness of hands $r = -0.505$ at $P = 0.001$). Thus, the clearer pronounced the mutation, the earlier developed speech disorders and coordination of fine motor functions in SCA1. In the final stage, speech impairment almost went to complete aphasia (out of 41 patients in 5 patients), combined with pronounced violations of the coordination of limb movements, ocular musculature and swallowing. So, the SF level of the SCA1 patients from a significant restriction of communication with one person (contact with relatives) to the complete cessation of communication with the public environment directly influenced their ability to attract people's attention ($r = +0.481$ at $P = 0.001$) and the possibility of assistance from neighbors ($r = +0.593$ at $P = 0.001$). At the same time, SF of the SCA1 patients is directly dependent ($r = +0.739$ with a confidence of 0.001 Pearson) from the RE scale (see below).

This Role-Emotional Role scale (RE), having impact on the level of performance of work or other daily activities (including a large time expenditure, a decrease in the amount of work, a decrease in its quality, etc.) of the SCA1 patients was 29.04 ± 5.76 points. The index of this control group was 1/3 (97.92 points) and was slightly less than 1/2 in the city group (70 points [7]). The value of the RE scale of the SCA1 patients also directly depended on the PF and RP scales ($r = +0.63$ and $+0.531$ with $P = 0.001$ by Pearson) and with VT as well (at $r = +0.683$) and was correlated with SF, and mental health MH ($r = +0.829$ at $P = 0.001$ by Pearson).

The scale "Mental Health - MH QOL" of the SCA1 patients, which characterizes the mood background (presence or absence of depression, anxiety) and emotional state of the SCA1 patients, was 1.31 ± 0.80 points. In the control group MH was 93.70 points, and among the inhabitants of Yakutsk - 67 points [7]. The revealed features of mental health of the SCA1 patients reflected its poor health in the patients affected by PF and RP (at $r = +0.510$ by Pearson with $P = 0.05$ and 0.001).

At the same time, the integral Physical Health Component (PH) of QOL of the SCA1 patients was 39.0410 ± 1.42495 points. Thus, the physical component of the health of the SCA1 patients was 74.22% of the number of the residents not suffering from SCA1 (52.68 points according to A.E. Mikhailova [7]). And the integral psychological component of health (Mental Health - MH) of QOL of the SCA1 patients was 24.4153 ± 1.41797 points. And the mental component of the health of the patients with SCA1 was 45.37% of the value of the health component of the non-SCA1 people (53.8 points [7]).

Along with these characteristics of the lowering of QOL of the SCA1patients there were groups, established among them: those who did not have close relatives (12.9%), those with disoriented attention (7.7%) and 5.1% of those who "did not count on or sought the aid of neighbors who with very great difficulty or just with difficulty helped others around them" (5.4%). Moreover the number of people, who received help of neighbors, directly depended on the number of people, who paid attention on the patients ($r = +0.676$ at $P = 0.001$). The number of close people depended inversely on the number of people who paid attention ($r = -0.415$ at $P = 0.05$ according to Spirman). The last indicator was in inverse relationship ($r = -0.522$ at $P = 0.001$) with the frequency of CAG repeats.

All the above mentioned showed that the SCA1patients with strong speech disorders experienced both more difficulties in communicating and establishing communication with others. At the same time, the number of psychological health problems such as anxiety ($58.5 \pm 0.78\%$) and depressive state ($53.7 \pm 0.778\%$) corresponded to the MH index of 1.31 ± 0.80 of the SCA1 patients. This means that patients with severe communication and psychological problems need timely specialized medical care and social assistance. Although the fact is complicated by the living of the SCA1patients in remote communities with transport problems, the problems still can be solved by the authorities of the Republic.

CONCLUSION

For the first time, an integrated assessment of QOL of the SCA1patients using the SF-36 Questionnaire showed a sharp decrease in QOL due to a sharp restriction of physical, role motor functioning, combined with impaired communicative functions and mental unhappiness that occurred among more than half of the observed SCA1 patients. SCA1patients with such characteristics should be maintained as a high-risk group.

This section of research was conducted for the first time in Yakutia as a center of accumulation of SCA1 by the method of refining the parameters of QOL of such patients. The negative nature of the established changes in QOL of the SCA1patients is related to the level of the mutations that have arisen, representing a scientific reserve, the continuation of which will have a practical solution as an opening of the social and psychological features of the SCA1clinic. The data obtained can be used in possible recommendations for the assistance and treatment of SCA1patients and will help specialists who choose the means of rehabilitation and maintenance of SCA1patients at home, as well as doc-

tors who determine the profile of institutions where established high-risk group patients are sent.

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REFERENCES

1. Tihonov D. G., Gol'dfarb L.G., Neustroeva T.S. Jakovleva N.V., Timofeev L.F., Luckan I.P., Platonov F.A. Analiz prodolzhitel'nosti zhizni i smertnosti bol'nyh spinocerebelljarnoi ataksiei 1 tipa [The analysis of the lifespan and mortality of patients with spinocerebellar ataxia type 1] Problemy social'noi gigieny, zdavooohranenija i istorii mediciny [The Social Hygiene, Public Health and Medicine History Problems]. 2015, №6, p. 29-31.
2. Ahmedova O.S. Psihologicheskii status i kachestvo zhizni pacientov s hronicheskoi obstruktivnoi bolezn'yu legkih [The psychological condition and the quality of life of patients with chronic obstructive pulmonary disease]. Avtoref. dis. kand. psiholog. nauk [PhD on psychol. Thesis]. Sankt-Peterburg, 2008.
3. Bivalcev V.A., Belov E.G., Sorokovnikov V. A. Ob ispol'zovanii ankety so shkalami boli vertebrologii [About the using the questionnaire scales for ache in vertebrology // Zhurnal nevrologii i psichiatrii [Journal neurology and psychiatry]. 2011, V.9. vyp.2, p.51-54.
4. Belyalova N.S. Instrukcija po obrabotke dannyh, poluchennyh s pomosh'yu oprosnika SF-36 [Instruction for calculation of the materials quality of life according its questionnaire SF-36] Googl.crom.ru/therapy.irkutsk./ doc/sf 36a.
5. Mingazova E.Z. Kliniko - yepidemiologicheskaja i molekuljarno-geneticheskaja izuchenie progressivnyh spinocerebelljarnykh ataksii v Respublike Bashkorstan [Clinical-epidemiologic and molecular-genetic study of progressive spinocerebellar ataxia in Bashkorstan Republic]. Avtoref. dis. kand. med. nauk. [The author's abstract of the dissertation of the candidate of medical sciences]. Moscow, 2009.
6. Mikhailova A.E. Klinichesk- ie osobennosti, kachestvo zhizni i farmakoyekonomicheskie aspekty terapii bol'nyh osteoartrozom v g. Jakutsk [Clinical particulars, quality of life and pharmaco-economical aspects of therapy osteoarthroses in Yakutsk] Avtoref. dis. na soisk. kand. med. nauk [The author's abstract of the dissertation of the candidate of medical sciences]. Moscow, 2006.
7. Movchan E.A., Pikalova N.N., Tov N.L. Kompleksnaja harakteristika kachestva zhizni bol'nyh na gemodialize v novosibirskoi oblasti [The complex characteristic of quality of life of patients on hemodialysis in Novosibirsk region] Zhurnal «Medicinskoe obrazovanie v Sibiri» Razdel medicinskie nauki 2012 g. № 1 / setevoe izdanie NGMI [Journal "Medical education in Siberia" Section of medical Sciences 2012 № 1] / the network edition of NGMI / ngmi.ru/coso/mos/article/pdf.
8. Nedoshivin, A. A., Kutuzov A. E., Petrov N. N. Issledovanie kachestva zhizni i psihologicheskogo statusa bol'nyh s hronicheskoi serdechnoi nedostatochnost'yu [The study of quality of life and psychological status of patients with chronic heart failure] Serdechnaja nedostatochnost' [Heart failure], volume 1, №4, 2000.
9. Neustroeva T. S., Platonov F. A., Tikhonov D. G. Narushenija social'nykh kontaktov u bol'nyh spinocerebelljarnoi ataksiei 1 tipa [Violations of social contacts in patients with spinocerebellar ataxia type 1] Tezisy dokladov V-i Mezhdunarodnoi nauchno-prakticheskoi konferencii, posvjashhennoi 95-letiyu P.A. Petrova «Problemy vilyuiskogo yencefalomielita i drugih neurodegenerativnykh zabolevanii: sovremennye voprosy yetiologii i patogen-eza» [Abstracts of Vth International scientific-practical conference dedicated to the 95th anniversary of P. A. Petrov "The problem of the Vilyuisk encephalomyelitis and other neurodegenerative diseases: current issues of etiology and pathogenesis"]. Yakutsk. June 23-24, 2016. Yakutsk: Publishing information centre, Alaas. 2016. P. 68-71.
10. Novik A. A., Ionova T. I. Rukovodstvo po issledovaniyu kachestva zhizni v medicine [Guide to the study of quality of life in medicine]. Sankt-Peterburg, 2002, P.37-315.
11. Platonov F. A. Spinocerebelljarnaja ataksija v Jakutii [Spinocerebellar ataxia in Yakutia] Avtoref. diss. dokt.med. nauk [The author's abstract of the dissertation of the doctor of medical sciences]. Moscow, 2003, 29 p.
12. Pogossova N.V. Mayak H. I., Yuferova, Yu. M. Kachestvo zhizni bol'nyh s serdechno-sosudistymi zabolevanijami: sovremennoe sostojanie problemy [Quality of life in patients with cardiovascular diseases: current status of the problem]. Cardiology.2010. room 4/ HTML version of the document from 23.11.2015
13. Iwasaki Yasushi, Mori Keiko, Ito Masumi. Presenile onset of spinocerebellar ataxia type 1 presenting with conspicuous psychiatric symptoms and widespread anti-expanded polyglutamine antibody- and fused in sarcoma antibody-immunopositive pathology//Psychoger-

atrie.Sep.2015. vol.15. p. 213-217.

14. F.A. Platonov, K. Tyryshkin, D.G.Tickonov, T.S. Neustroyeva, N.V. Yakovleva, T.M. Sivtseva, O.G.Sidorova, V.P. Nikolaev, L.G. Goldfarb, N. M. Renwick. F. A. Genetic fitness and selection intensity in a population affected with high-incidence spinocerebellar ataxia type 1. // Neurogenetics, 2016, 17: 179-185, DOI 10-1007/s10048-016-0481-5.

15. Olivopontos cerebellar atrophy in large Yakut kinship in Eastern Siberia. L.G. Golfard, M.P. Chumakov, P.A. Petrov [L.G. Golfard, M.P. Chumakov, P.A. Petrov, N.I. Fedorova, D.C. Gajdusek] // Nevrology. 1989, vol.39, n.11, p. 1527-1530

16. [Rakowicz M.](#), [Sulek A.](#), [Sobanska A.](#) Prospective study of motor cortex excitability and conduction of pyramidal tracts in the presymptomatic spinocerebellar ataxia type 1 gene carriers // [Journal](#)

[of the Neurological Sciences](#). 2015. Oct. Supplement 1, Vol. 357, p. 284-e284.

17. Ware J.E., Kosinski M., Keller S.D. SF-36 Physical and Mental Health Summary Scales: A User's Manual // The Health Institute, New England Medical Center. Boston, Mass.-1994.

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ANALYSIS OF ASSOCIATIONS OF GENETIC POLYMORPHISMS 481C> T, 590G> A I 857G> A GENE OF THE ENZYME N-ACETYLTRANSFERASE 2 (NAT2) WITH THE RISK OF LUNG CANCER IN THE YAKUTS

ABSTRACT

For the first time the search of polymorphic options of a gene of NAT2 associated with the development of lung cancer in Yakutia has been carried out. Genetic markers of the increased and lowered risk of development of lung cancer in the Yakuts have been revealed. It is established that markers of the increased risk of development of lung cancer for the Yakuts are the allele of NAT2*857A and a genotype of NAT2*857G/A, markers of the lowered risk – NAT2*857G allele, NAT2*857G/G genotype.

Keywords: lung cancer, polymorphic options, N-acetyltransferase-2. **Keywords:** lung cancer, polymorphic options, N-acetyltransferase-2.

INTRODUCTION

In Russia cancer of a lung takes the leading positions in structure of oncological incidence – the incidence of it for the last 20 years has increased more than twice and it is on the first place among malignant neoplasms [5]. Annually in Russia this pathology is diagnosed more, than for 63 thousand patients. The problem of cancer of lung is relevant also for Yakutia where this form of cancer for many years takes the first place in structure of an oncopathology. In Republic Sakha (Yakutia) with the population in 982,1 thousand people annually more than 300 people get cancer of a lung [2]. The sharpness of a problem is caused not only by high prevalence of a disease, but also by late diagnostics as at an early stage lung cancer is possible

to diagnose no more than in 15% of cases [4]. It is relevant to study all factors involved in carcinogenesis.

Lung cancer, as well as many oncological diseases, is a multiple-factor disease and in its development an important role is played as outside environment (smoking, asbestos, radon, arsenic, etc.), and genetic factors [7, 9, 11, 17]. By some authors, it is shown that polymorphic options of a gene of NAT2 make a contribution to development of oncological diseases including cancer of a lung [8, 13, 18].

The gene of NAT2 is localized on a short chromosome arm 8 (8p23.1), has about 9900 PN extension, contains 2 exons and it is expressed mainly in a liver and intestines [3, 14]. The N enzyme-acetyltransferase-2 coded by this gene

represents the protein with a molecular weight of 33 kd consisting of 290 amino-acid residues. This enzyme localized in cytoplasm participates in process of biotransformation of aromatic amines, which are present at the environment. A source of aromatic amines are industrial wastes, pollution of water, air, and a number of medicines [3, 15].

MATERIAL AND METHODS

In the present article it has been done a comparative studying of polymorphism of a gene of NAT2 enzyme arylamine N-acetyltransferase among patients with cancer of a lung and among healthy people, residents of the Sakha (Yakutia) Republic. We have examined 60 patients with cancer of a lung of Yakut ethnic origin from which 43 men, 17 women received treatment in a republican oncological