(Per 100 thousand population) [4-6]

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EPIDEMIOLOGY OF PARKINSON'S DISEASE IN THE RS (YA)

ABSTRACT

Parkinson's disease (PD) is the second most common neurodegenerative disorder in the world after Alzheimer's disease. The prevalence of the disease varies widely in different ethnic and geographical groups. The purpose of the research is to study the epidemiological picture of PD in the population of the Sakha (Yakutia) Republic. We used the next sources of information: 1) own data collected during the examination of patients; 2) retrospective analysis of medical records about patients of the neurological department; 3) data from annual reports of neurologists; 4) the results of selective population studies conducted by the Department of neurology and psychiatry of Medical Institute of NEFU. All information was entered in the database «Register of patients with Parkinson's disease in the Sakha (Yakutia) Republic». The prevalence of PD in Yakutia was 67 per 100,000 of the adult population. The disease was more common in women than in men: 79.7 versus 52.9 per 100,000. The prevalence of the disease in Yakutsk was 76.5 per 100,000 population, and in the regions it varied widely from 9.8 to 185.6 per 100,000 population. The highest frequency of PD was found in the central regions (Gorny, Khangalassky), in the regions of the Vilyui group (Vilyuisky and Verkhnevilyuisky), as well as in Tattinsky, Abyisky, Verkhnekolymsky and Lensky regions. The prevalence of PD increased in the older age groups: in the 40-49 age group this parameter was 12.9 per 100,000, and in the 70 years and older group it was 527.5 per 100,000. We did not reveal statistical differences in prevalence of PD among the Yakut (75.4 per 100,000) and Russian (73.4 per 100,000) population. The incidence of PD persists at a low level, but over the past 5 years, there has been a tendency to increase it: if in 2011 it was 1.42 per 100,000, in 2016 it would be 3.86 per 100,000 population. Thus, we conducted an epidemiological study of PD in Yakutia for the first time, identified regional features, found that the disease with the same frequency occurs in the Yakut and Russian population.

Key words: Parkinson's disease; epidemiology; prevalence; incidence.

INTRODUCTION

Parkinson's disease (PD) is one of the most common age-dependent neurodegenerative diseases [4]. If PD is extremely rare up to 40 years, among those over 60 years of age prevalence reaches 1%, and among those over 80 years old - 4% [8].

The epidemiological data of the disease varies widely in different ethnic and geographical groups [6]. For example, prevalence of PD in France is 308 [9],

Great Britain – 128 [12], and Egypt – 436 per 100,000 population [10]. In Russia, the epidemiological situation of PD is heterogeneous, and prevalence rates range from 17 to 139.9 per 100,000 population [5, 7]. This situation is probably due to different designs of the studies conducted, as well as the hypodiagnosis of the disease itself due to low awareness of the population, the difficulties in differential diagnosis with other motor disorders accompanied by parkinsonism [3, 5].

Therefore, epidemiological studies of PD is important for determining potential risk factors and improving understanding of the course of the disorder. In addition, these data are used for effective planning of medical care and rational use of health resources [2, 11].

The purpose of the research is to study the epidemiology of PD in the population of the Sakha (Yakutia) Republic.

MATERIAL AND METHODS

The study was conducted at the Department of neurology and psychiatry of the Medical Institute of M.K. Ammosov North-Eastern federal university (NEFU) and the clinical bases of this department: the neurological department of Republican Hospital Number 2 – The Center of emergency medical aid, University Clinic of NEFU in the period from 2015 to 2017. The study was approved at the meeting of the Local Committee for Biomedical Ethics of the Yakutsk Scientific Center for Complex Medical Problems (Protocol No. 43 of 9 November 2016, Decision No. 2).

We used the next sources of information: 1) own data collected during the examination of patients; 2) retrospective analysis of medical records about patients of the neurological department; 3) data from annual reports of neurologists; 4) the results of selective population studies conducted by the Department of neurology and psychiatry of Medical Institute of NEFU. All information was entered in the database "Register of patients with Parkinson's disease in the Sakha (Yakutia) Republic".

At the end of the research period, following indicators were calculated: prevalence – the total number of patients with PD in the population, calculated for 100,000 of the population; incidence – the number of new cases of PD in the population for 1 year, calculated for 100,000 of the population.

Epidemiological data were calculated for the adult population of Yakutia, for individual regions, and for males and females. The prevalence was calculated among the Russian and Yakut population.

RESULTS

The population of the Yakutia at the time of data processing (March 2017) was 959,689 people, including persons of 18 years and over - 698,735 people, persons 40 years and over - 371,637 people. During the period of the study in the Yakutia, there were 468 cases of PD. The prevalence of the disease was 67 (95% CI: 60.9-73.0) per 100,000 of the adult population, which is almost 2 times lower than the global figure.

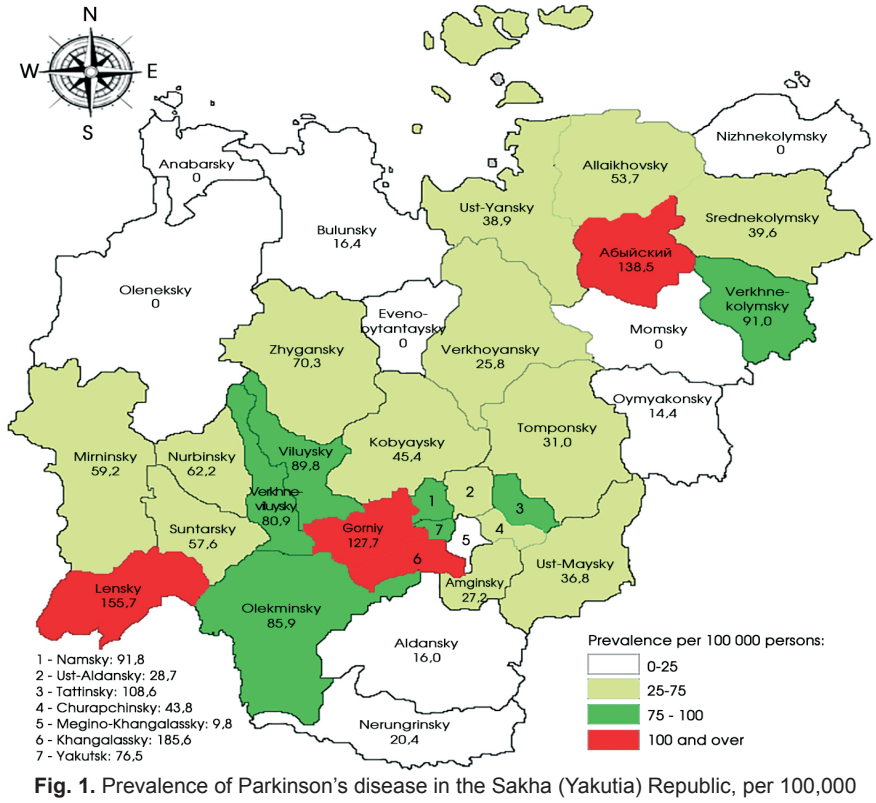

Fig. 1. Prevalence of Parkinson's disease in the Sakha (Yakutia) Republic, per 100,000 population

PD was more common in women [79.7 (95% CI: 80.2-100.9) per 100,000] than in men [52.9 (95% CI 40.9-55.1) per 100,000], which can be explained by the greater concern of women with their state of health, as well as by the shorter life expectancy of males. In general, the ratio of men and women was 1: 1.65.

PD was identified in 29 of the 34 administrative regions of the Yakutia and in Yakutsk. The prevalence of the disease in the capital of the republic, in the city of Yakutsk, was 76.5 (95% CI: 65.7-87.4) per 100,000 people, and in the regions varied from 9.8 to 185.6 per 100,000 (fig. 1).

The highest frequency of PD was found in the central regions (Gorny, Khangalassky), in the regions of the Vilyui group (Vilyuisky and Verkhnevilyuisky), as well as in Tattinsky, Abyisky, Verkhnekolymsky and Lensky regions. In this regions of the Yakutia prevalence of PD coincide with global indicators. Lower prevalence levels were recorded in the northern regions: Ust-Yansky, Bulunsky, Srednekolymsky, Verkhoyansky and others, and in the two southern regions – Aldansky and Neryungrinsky.

We have identified areas that are "free" from PD: Anabarsky, Momsky, Nizhnekolymsky, Oleneksky, Eveno-Bytantaisky. The absence of PD cases in these administrative-territorial regions can be explained by insufficient specialized care for patients with neurological diseases. However, this fact is probably also due to a low population, including elderly and senile patients and, correspondingly, a smaller expected number of patients with PD.

A study of the prevalence of PD among the age groups showed that the frequency of occurrence of the disease increases in older age groups: in the 40-49 age group the prevalence was 12.9 (95% CI: 6.6-19.3) per 100,000 population, in group of 70 years and older – 527.5 (95% CI: 454.4-600.6) per 100,000 population (Fig. 2).

The study showed that the prevalence of PD in women is higher in all age categories, except for the group of 70 years and older, where this parameter for men was 560.4 (95% CI: 427.6-693.2) per 100,000, for women – 511.9 (95% CI: 424.5-511.9) per 100,000 (Fig. 3).

According to the results of the All-Russian Population Census in 2010 in the Yakutia lived 958,528 people, of which 466,492 –Yakut ethnic group and 353,649 - Russian [1]. We have standardized the population by ethnic group for the adult population of the republic for 2017 to compare the frequency of occurrence of PD. As a result, it was revealed that the disease was spread with practically the same frequency among the Yakut ethnic group and among the Russian population. Thus, this indicator was 75.4 (95% CI: 66.2-84.6) per 100,000 for the Yakut population and 73.4 (95% CI: 62.9-83.9) per 100,000 for the Russian population.

The incidence of PD in 2011 was 1.42 per 100,000 of the population. Within 5 years there has been a steady increase in this indicator, which reflects an improvement in the diagnosis of the disease. So, in 2016 the incidence of PD was 3.86 per 100,000

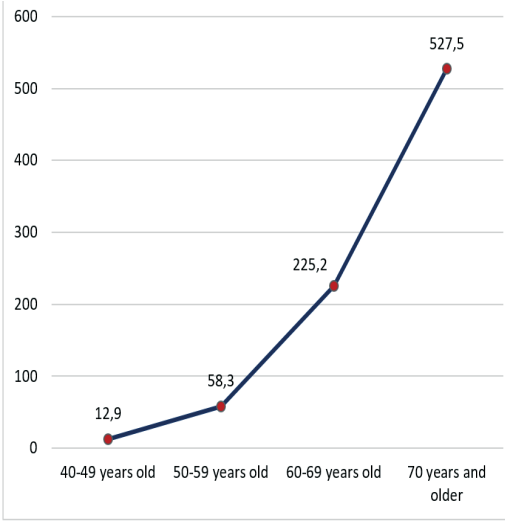

Fig. 2. revalence of PD in different age groups, per 100,000 population

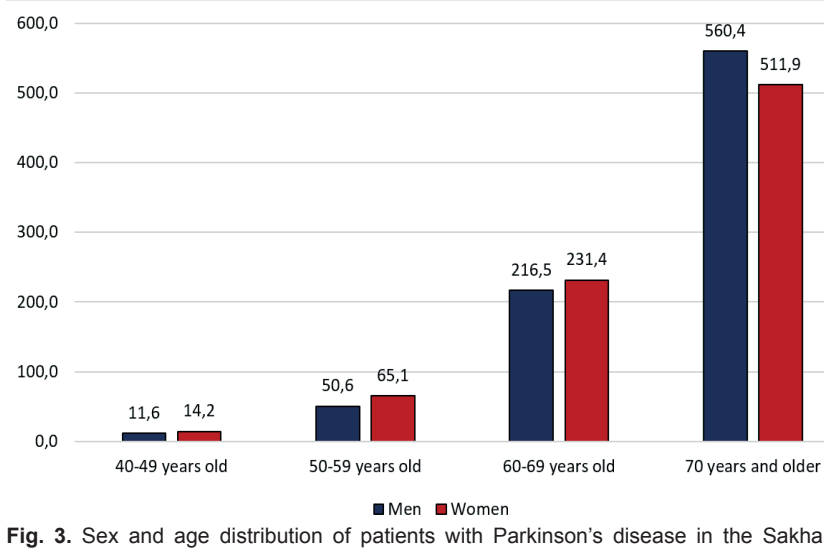

Fig. 3. Sex and age distribution of patients with Parkinson's disease in the Sakha (Yakutia) Republic

The incidence of Parkinson's disease in the Sakha (Yakutia) Republic in 2011-2016, per 100,000 of the population

Year	2011	2012	2013	2014	2015	2016
Incidence	1.42	1.25	1.39	2.29	2.58	3.86

of the population (Table 1). However, the indicator does not reach the international level, which requires further improvement of the quality of care in this category of patients.

CONCLUSION

The prevalence of PD in the Yakutia is heterogeneous. The disease is more evident in those rural areas where the work of a neurologist is well established. The prevalence of patients with female PD is due to women's concern for their health and longer life expectancy. The wide gap between epidemiological indicators in the neighboring areas indicates the role of medical support in the active detection of PD. In addition, in some parts of the region, especially in northern areas, cases of PD are not identified, which may be explained by the small number of elderly and senile patients and the low expected number of patients. The differences in frequency of PD among the Yakut and Russian population were not found. The increase in the incidence of PD in the last 5 years is associated with the appearance of a parkinsonologist in the Yakutia.

Thus, in order to improve the organization and increase the availability of specialized medical care for patients with PD, the organization of a educational base for neurologists, the introduction of new methods of diagnosis and treatment in practice, improving the social adaptation of patients in the Sakha (Yakutia) Republic, there is a need to create a center for extrapyramidal pathology.

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DYNAMICS OF THE STRUCTURE OF GASTROENTEROLOGICAL MORBIDITY IN PATIENTS OF DISPENSARY GROUPS OF 20- YEAR OBSERVATION

ABSTRACT

The dynamics of the structure of gastroenterological morbidity is analyzed on the example of 134 patients p. Vilyuchan of the Suntarsky district, who were under clinical observation for 20 years since 1980 and were subjected to repeated examinations (in 1990, 1997 and in 2001).

Keywords: Disease, digestive organs, structure of morbidity, dispensary groups, dynamics.

MATERIALS AND METHODS

Prophylactic medical examination in 1980 passed 422 people (171 men and 251 women), with a total adult population of 495 people, or 85.25% of the population. According to the size of the population p. Vilyuchan, by the time of the second inspection in 1990, out of the first examined, 189 people had left, 135 out of them in connection with the departure and 54 due to death. In 1990, with an adult population of 484 people, 77.3% were examined and 233 people were examined again. More than half of the contingent was people in Age from 18 to 40 years. Individuals aged 60 years and over were 17.6%.

In 2001, 69.19% of the 422 adults surveyed for the out-of-hospital population were examined, including 134 patients registered on dispensary since 1980. According to the data received, in 1980, 76 people were practically healthy (18% of those surveyed), in 1990 - 14 (3.7%), of the 134 people in 2001, only 6 people were practically healthy (4, 48%).

RESULTS AND DISCUSSION

General incidence of adult population p. Vilyuchan of the Suntar region is characterized by high indicators both in 1980 and 1990 and has a tendency to increase in subsequent years.

In the structure of the general incidence of digestive diseases, the leading place is occupied by the leading one. In 2nd place are diseases of the blood circulation system, in 3rd place - in 1980 and 1990 diseases of the respiratory system.

In 2001, the third place went to diseases of the genito-urinary organs (Fig. 1). At the 4-th place in the structure of the general incidence of the adult population - diseases of the musculoskeletal system and connective tissue.

In the dynamics of medical examination for 1980-1990-2001, the growth rates of the incidence of the population is observed in many classes of diseases: the circulatory system, digestive system, musculoskeletal system and connective tissue, nervous system, urogenital organs due to urolithiasis and uric acid Diathesis. Especially it should be noted the growth of helminthiases.

Those who were practically healthy during the preventive examination were 180.1 ‰ in 1980, 37.7 ‰ in 1990, 20.54 ‰ in 2001

Analysis of gastroenterological morbidity in the population p. Vilyuchan, according to the data of preventive examinations conducted in 1980-1990-2001, shows a high prevalence of diseases of the digestive organs and their non-growth in dynamics.

The most common diseases of the stomach and duodenum, although in recent years, their decrease in morbidity and specific gravity (Fig. 2).

At the same time there was a statistically significant increase in the diseases of the hepatobiliary system and pancreas and their sharp growth by 2001. It is alarming the sharp growth of calculous and calculous cholecystitis in 1990 with a 4-fold increase in the number of patients with postcholecystectomy syndrome in 2001 and giardiasis cholecystitis. The prevalence of colon diseases continues to be high in 2001, but the highest incidence rates of colon diseases, exceeding the 1980 data by 5.7 times, were noted in 1990 (Fig. 3).

In p. Vilyuchan in the course of 20 years, was followed by 134 people undergoing medical check-up, both in 1990 and in 2001, of which 103 were examined in 1997.

44 patients out of 103 gastroenterological patients, examined in 1997, in 1980 were practically healthy, 11 of which were in those years in childhood and adolescence. By age - these are people who by 2001 have become older (compared to 1980) for 20 years.

In the structure of morbidity the pathology of the digestive organs predominates with a consistent increase in indices. Invariably the 2nd place is occupied by diseases of the circulatory system. At the same time, the number of diseases of the circulatory system has increased.

Of particular note is the sharp increase in diseases of the genitourinary system (urine-stone disease and urine acid diathesis), the musculoskeletal system and connective tissue (osteochondrosis), the nervous system (posttraumatic and discirculatory encephalopathy). Among gastroenterological patients, 9 people suffered from obesity. Attention is drawn to the subsequent growth of cases of tuberculosis of respiratory organs, as well as gynecological diseases, mainly

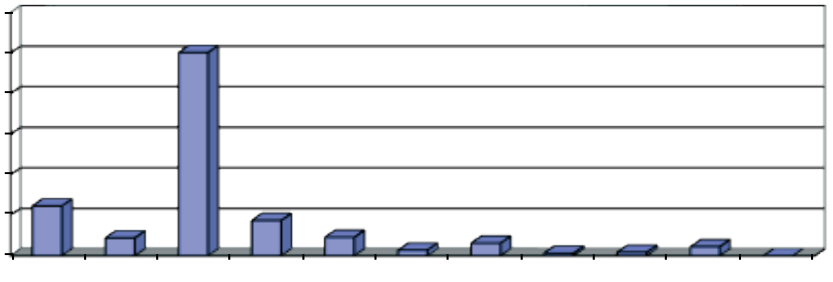

Fig. 1. Morbidity of the population p. Vilyuchan of the Suntarsky district according to the data of the medical check-up of 2001.

1 - diseases of the circulatory system; 2 - diseases of the respiratory system; 3 - diseases of the digestive system; 4- diseases of the urino-genital organs; 5 - diseases of the musculoskeletal system and connective tissue; 6 - diseases of the endocrine system; 7 - diseases of blood and blood-forming organs (anemia); 8 - diseases of the nervous system; 9 - tuberculosis of the respiratory system; 10 - gynecological diseases; 11 - other.

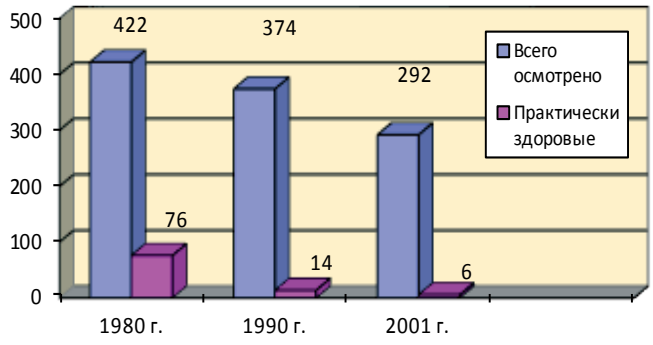

Fig.2. The contingent of practically healthy according to the data of professional examinations in dynamics.
Those who were practically healthy during the preventive examination were 180.1 ‰ in 1980, 37.7 ‰ in 1990, 20.54 ‰ in 2001

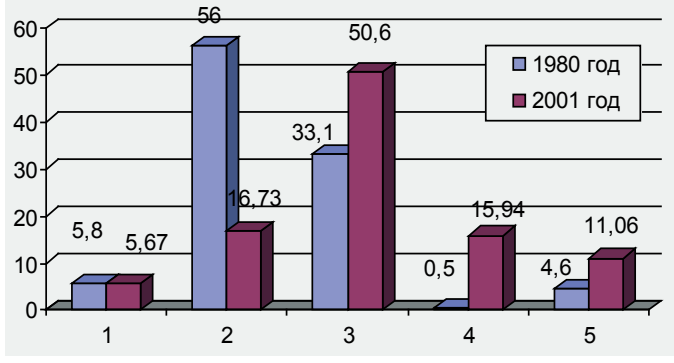

Fig. 3. Gastroenterological morbidity of the population p. Vilyuchany
1 - diseases of the esophagus; 2 - diseases of the stomach and duodenum; 3 - diseases of the hepatobiliary system; 4 - pancreas diseases (pancreatitis); 5- Bowel disease.

due to uterine myoma. In the described period, there was an increase in the incidence of diseases of food-digestive organs (Fig. 4).

The highest rates of growth in morbidity rates are characteristic for diseases of the hepatobiliary system, pancreas and intestine, mainly due to colonopathy. At the last dispensary examination in 2001, 68 patients from 103 patients of the previous examination of 1997 passed a gastroenterological examination. Among them were 28 men and 40 women. In the dynamics, the increase in the diseases of the gastrointestinal tract was revealed from 237 cases in 1997 to 293 cases in 2001, mainly in women (from 143 to 181 cases, respectively).

In the structure of gastroenterological morbidity there is a decrease in the specific gravity of diseases of the esophagus, stomach, duodenum and stomach with an increase in the diseases of the hepatobiliary system and the pancreas (Fig. 5).

At the same absolute absolute number of patients with esophageal pathology, in the compared years there was an increase in men of reflux esophagitis II and III degree of severity. A high percentage of chronic gastritis with hypoxecretion and achlorhydria in recent decades can be related to the age of patients who, by the time of the last two examinations, are older by 17-20 years.

The pattern of changes in the gastric mucosa, confirmed by morpho-endoscopic studies, was traced in 29 healthy and 40 patients with chronic gastritis, examined in dynamics at intervals of 20, 10, 7 years and 3 years. In dynamics, the growth of pathomorphological changes in the gastric mucosa as in practically healthy patients and in patients with superficial gastritis is clearly traced. Already in 1990, only one woman was found to be practically healthy, the rest of the patients, who were healthy at the first examination, had gastric changes in the gastric mucosa. The aggravation of the degregenerative de-structural changes in the gastric mucosa with a persistent decrease in the level of acid formation was noted in 28 of 40 patients with chronic gastritis.

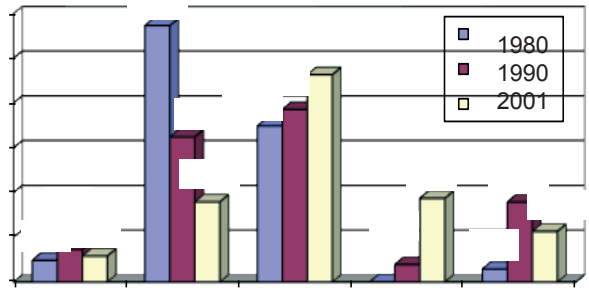

Fig. 4. Structure of gastroenterological morbidity in dynamics
1 - diseases of the esophagus; 2- diseases of the stomach and duodenum; 3- diseases of the hepatobiliary system; 4 diseases of the pancreas; 5 - bowel diseases.

The esophagogastroduodenoscopy, carried out from 1980 to 2001 in dynamics 4 times, indicates the involvement of the esophagus, duodenum, hepatobiliary system in the pathological process, as

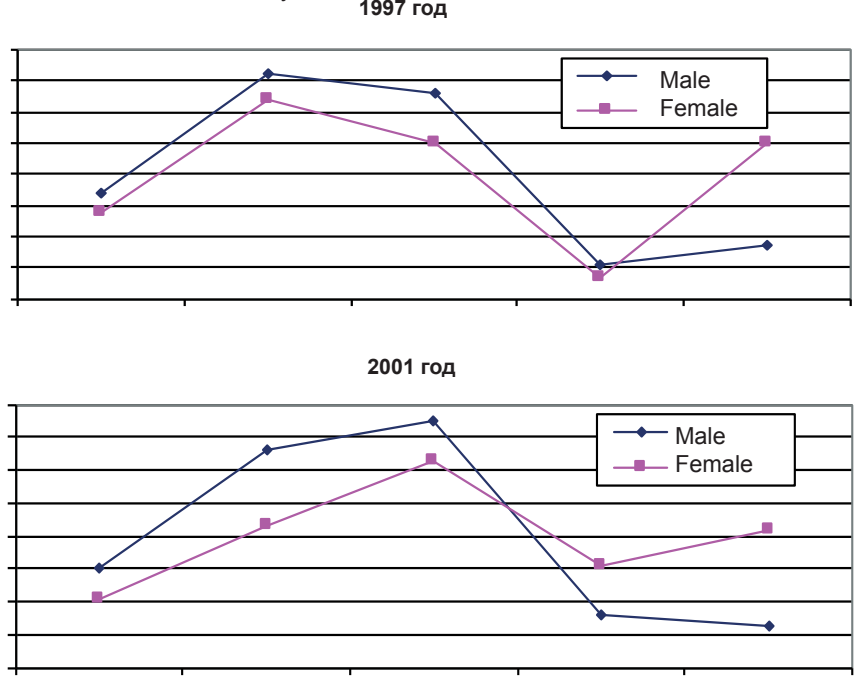

Fig. 5. Structure of gastroenterological morbidity
1 - diseases of the esophagus; 2- diseases of the stomach and duodenum; 3- diseases of the hepatobiliary system; 4 diseases of the pancreas; 5 - intestinal diseases.

well as the violation of the motor and evacuation function of the gastrointestinal tract.

Perennial endoscopic observations of practically healthy individuals and patients with various forms of chronic gastritis confirm the progression of changes in the gastric mucosa with the age of the patient and the absence of reverse development of atrophic gastritis. In patients with chronic gastritis, regardless of its clinical morphological variant, stratification of the pathology of other organs of the digestive tract aggravates the course of the underlying disease.

Among the accompanying pathology in gastroenterological patients, the leading place in 1997 was occupied by diseases of the circulatory system, exceeding the primary index by 5 times in both men and women (Table 2.4.8). Continuing to increase, the diseases of the circulatory system receded to the third place, giving way to diseases of the genitourinary organs and the musculoskeletal system. The frequency of detection of respiratory diseases in gastroenterological patients in the last decade (according to 1997 and 2001) moved to the 4-th and 6-th places. Women with age have a higher proportion of diseases of the endocrine system and anemia (Table 2.4.9).

Thus, the frequency of concomitant pathology is determined by the age and sex of the patient.

Attention is drawn to the intensive growth of osteochondrosis with the age of the patients: in men 6 times, in women 23 times. Compared with baseline data, the frequency of diseases of the musculoskeletal system exceeded in 2001 in men 10 times, in women 45 times. There is concern about the continuing increase in the incidence of urolithiasis, whose incidence in 2001 was many times higher than the primary rates. It is alarming the growth of calculous (from 0 to 20 cases) and lambliasis cholecystitis (from 0 to 15 cases).

In the structure of gastroenterological morbidity, the leading role belongs to the diseases of the hepatobiliary system, pancreas and intestines. Dynamic examination with an interval of 10 years allows to assess the degree of progression of the degregenerative and dystrophic processes of the gastric mucosa, the risk of transformation into more severe forms of chronic gastritis, the layering of complications of the esophagus and intestine.

The effect of various exogenous and endogenous factors on the development of chronic processes was traced in 102 patients with gastritis with a decreased secretion, examined in dynamics based on anamnestic data analysis for 12 factors. The average age of men was 49.04 ± 0.95 years, women - 50.92 ± 1.69 years. For 10 years (1980-1990), with a preserved average growth of 154.2 ± 0.9 cm, body weight increased from 56 ± 0.01 to 58.7 ± 0.97 kg.

Hereditary complication in oncological diseases was detected in 33 patients (32.3%) with a distinct predominance of weighting along the line of the father (19.6 ± 3.91%), the hereditary maternal burden was noted in 8.82 ± 8, 1% of patients, in the line of both parents and direct relatives - 0.98 ± 0.97 and 2.94 ± 1.67%, respectively.

Of the 102 patients, most do not smoke, including those who quit smoking on the recommendation of doctors in connection with the disease (59.8 ± 4.85%), 38.24 ± 4.81% of patients are constantly smoking at least a pack a day. Absolutely do not drink alcoholic beverages 16.67 ± 3.19% of patients, rarely consuming were 54.9 ± 4.92%. Alcohol consumption once a month was noted in 7.84 ± 2. 66% of patients who regularly drink once a week - 8.82 ± 2.81%, threw out drinking for various reasons was 9 people (8.82 ± 2.8%).

One of the risk factors for the disease of the digestive system is tooth decay, noted in all patients examined in the dynamics. An essential role in the development of the disease of the digestive system has alimentary factors. In this case, a special place is occupied by a disturbance of the diet (62.75 ± 4.7%), consisting of irregular feeding (with long intervals between meals, overeating in the evening). A satisfactory quality of nutrition was noted in 92.16 ± 2.66% of patients, good - in 6.86 ± 2.5%, unsatisfactory nutrition was established in one. In the daily diet, meat of this group is dominated by meat (79.41 ± 4.0%), milk and dairy products (82.35 ± 3.77%), bread and flour products (90.2 ± 2.94%). The presence of fish in the grocery set is noted in 33.3 ± 6.6% of patients. Fresh vegetables in nutrition are seasonal in nature and their regular intake during the survey was detected in 8.82 ± 2.85% of patients. It should be noted a certain tendency to abuse sweets and fatty foods (8.82 ± 2.81 and 17.64 ± 3.77%, respectively).

Conjugation of diseases of the digestive system with the blood group is shown in 46 patients with chronic gastritis with a decreased secretion. Among the surveyed, the largest percentage were people with A (II) blood group (41.3%). Patients with 0 (I) and B (III) had 11 blood groups (23.9%), and A (IV) blood group was detected in 5 patients (10.9%).

The average age of patients with chronic gastritis with a decreased secretion corresponded to 50 years, which, perhaps, can be attributed to factors of cancer risk in connection with the hormonal changes and metabolic changes typical for this age period.

Based on the generalization of data, the above exogenous and endogenous factors should be considered as factors contributing to the emergence of the pathological process and its chronicization.

Long-term clinical and epidemiological studies conducted in the dynamics in p. Vilyuchan of the Suntarsky district of the Republic of Sakha (Yakutia), while increasing the level of general morbidity of adults, which shows a progressive deterioration in the health of the population. The obtained data are correlated with the statistical data of the M3 PC (H). For the period 1997-2001. There was an increase in the incidence of diseases of the hepatobiliary system, mainly due to diseases of the gallbladder and bile ducts. There is a tendency to rejuvenate cholelithiasis. It is connected, as it seems to us, not only with the improvement of diagnostics, but above all the true quantitative growth of the disease. Among the reasons that lead to the growth of gastrointestinal diseases, it is necessary to identify metabolic disorders, hypodynamia, lifestyle changes associated with social and economic transformation

Hypomotor dyskinesia of the bile ducts, which predominate among the indigenous population, regardless of their age, lead to the development of secondary inflammatory processes in the gallbladder. The growth of helminthiases and diseases of the gallbladder parasitic etiology is of particular concern.

Endoscopic studies with morphological verification, conducted in dynamics in practically healthy people and in patients with chronic gastritis, confirm the progression of pathomorphological changes in the gastric mucosa with the age of the patient, which must be taken into account when developing therapeutic and prophylactic measures.

Analysis of the acid-forming function of the gastric mucosa, carried out in dynamics on the same patients, shows the presence of interrelations of the secretory function of the stomach with the change in the state of the gastric mucosa with the age of patients.

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RESULTS OF NEONATAL SCREENING ON ADRENOGENITAL SYNDROME IN CHILDREN IN THE RS (YA) FOR 10 YEARS

ABSTRACT

The results of neonatal screening in the Republic of Sakha (Yakutia) (RS (Y)) and the endocrinology department of the Pediatric Center RB-1-NTSM for ten years on congenital dysfunction of the adrenal cortex (syn. Adrenogenital syndrome) are presented in the article. Adrenogenital syndrome is a group of diseases with an autosomal recessive type of inheritance, which is based on a defect of one of the enzymes or transport proteins involved in the biosynthesis of cortisol in the adrenal cortex. Neonatal screening for adrenogenital syndrome is an effective method of early diagnosis and treatment of a disease that can prevent the development of disabling complications and death. Coverage of newborns with neonatal screening in the RS (Y) annually increases. With timely treatment of adrenogenital syndrome, the rates of physical development and puberty of the child are approaching the norm. The prevalence of adrenogenital syndrome in the RS (Y) is lower than in the Russian Federation and its regions: in the Ural Federal District, in the Siberian Federal District. The most frequent occurrence of adrenogenital syndrome is observed in Alaska residents, the lowest in China. All patients have a lossy form of the disease. Analysis of patients identified by neonatal screening did not determine significant differences in gender, place of residence. In girls, the diagnosis was made immediately after birth due to the presence of virile syndrome. A case of an incorrect determination of the sex in a girl at birth was described on the patient's medical chart and was diagnosed with hypospadias. Substitution therapy for the majority was started up to 21 days. All patients receive replacement therapy with glucocorticoids and mineralocorticoids (Cortef, Cortineff) from the time of diagnosis in an individual dosage, depending on age. Acceleration of bone age is observed only in one child, in three, a decrease in the rate of growth and a lack of body weight. The organization of neonatal screening for adrenogenital syndrome in the RS (Y) allowed achieving a high percentage of the survey of newborns, reduction of the period of examination and early initiation of substitution therapy, prevention of disability of patients.

Keywords: congenital adrenal cortex dysfunction, neonatal screening, newborn, frequency.

INTRODUCTION

Congenital adrenal cortex dysfunction (ADHD, adrenogenital syndrome (AGS), congenital adrenal hyperplasia) is a group of diseases with an autosomal recessive type of inheritance, which is based on a defect of one of the enzymes or transport proteins that participate in the biosynthesis of cortisol in the adrenal cortex [2].

For the first time the disease was described by Phillips in 1886 as pseudohermaphroditism in a girl at the age of 19 days. In 1924 O.V. Vereshchinsky for the first time in the domestic literature cited information on 12 cases of adrenal-sexual syndrome. In the years 1950-1952. F.C.Bartter, F.Albright, A.Leaf, E. Dempsye, E. Carroll, L. Wilkins deciphered the essence of this disease, the biosynthesis of hydrocortisone. VDKN is the most common pathology of the adrenal glands in children (1 case per 5000 born).

Neonatal screening contributes to the early diagnosis of ACS, especially in boys before the development of clinical symptoms, to the early onset of substitution therapy and the safe social adaptation of children. Coverage of newborns with neonatal screening in the RS (Y) annually increases. With timely treatment of adrenogenital syndrome, the rates of physical development and puberty of the child are approaching the norm.

All patients have a lossy form of the disease. Analysis of patients identified by neonatal screening did not determine significant differences in gender, place of residence. In girls, the diagnosis was made immediately after birth due to the presence of virile syndrome. A case of an incorrect determination of the sex in a girl at birth was described on the patient's medical chart and was diagnosed with hypospadias. Substitution therapy for the majority was started up to 21 days. All patients receive replacement therapy with glucocorticoids and mineralocorticoids (Cortef, Cortineff) from the time of diagnosis in an individual dosage, depending on age. Acceleration of bone age is observed only in one child, in three, a decrease in the rate of growth and a lack of body weight.

MATERIALS AND METHODS

The order of the Ministry of Health of the Republic of Sakha (Yakutia) of March 20, 2006 was issued to organize screening, introduce new methods, organize diagnostic and therapeutic care. 01-8 / 4-134a «On the progress of the activities of the section of the national project» Health «on the examination of newborn children for hereditary diseases.» The Order of the Republic of Belarus No. 1-NCM dated August 31, 2006. №01-0108 / 91 «About rendering medical aid to children with cystic fibrosis, adrenogenital syndrome, galactosemia, phenylketonuria and congenital hypothyroidism, revealed by neonatal screening» [5]. The screening procedure includes blood sampling in full-term newborns on day 4 of life, in preterm patients on day 7 and determination of 17-hydroxyprogesterone (17-ONP) levels in samples using special screening kits. The level of 17-SNP in blood samples is determined by the immunofluorescence method (test kits «Delfia 17-α-OH Progesteron», Finland, and «17-α-OH-Progesterone-Immunoskrin», Russia). The following provisions are taken into account when interpreting the indicators of 17-SNP: - the level of 17-SNP for full-term children (the gestation period is more than 37 weeks, the body weight is more than 2000 gr.) Normally up to 30