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Wyzwania współczesnej medycyny – studium przypadku pacjenta z SLA po 60 roku życia

Challenges of modern medicine – a case study of a patient with ALS after age 60

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Key words: amyotrophic lateral sclerosis, case report, challenges of modern medicine

Słowa kluczowe: stwardnienie zanikowe boczne, stadium przypadku, wyzwania współczesnej medycyny

Abstrakt

Wstęp. Stwardnienie zanikowe boczne jest to choroba zwyrodnieniowa układu nerwowego, neurodegeneracyjna kory mózgowej, opuszki i rdzenia kręgowego na skutek uszkodzenia neuronów ruchowych górnego i dolnego, o postępującym przebiegu, różnych objawach ze zróżnicowanym natężeniem i czasem trwania (od kilku miesięcy do nawet 10 lat), o rokowaniu skrajnie niepomyślnym. Jako choroba nieuleczalna, postępująca, na skutek pogarszania sprawności ruchowej prowadzi do całkowitego zatrzymania pracy mięśni, w tym oddechowych, przy całkowicie zachowanej sprawności intelektualnej chorych

Opis przypadku. Pacjentka lat 65 hospitalizowana w Klinice Geriatrii Szpitala Uniwersyteckiego nr 1 im. A. Jurasza w Bydgoszczy z powodu zapalenia płuc. Chora z dusznością spoczynkową, (oddech przyśpieszony, spłycony) obecny paradoksalny ruch klatki piersiowej, osłabiona ruchomość klatki piersiowej, obecne furczenia, świsty w trakcie badania osłuchowego. W rozpoznaniu stwardnienie zanikowe boczne, przewlekła niewydolność oddechowa, nadciśnienie tętnicze, nadczynność tarczycy. W wywiadzie problemy z przełykaniem, ból jamy brzusznej, niewydolny odruch kaszlowy, dysfagia, podwyższona temperatura ciała, trudności z zaśnięciem, obniżony nastrój. Pacjentka nie wyraziła zgody na wykonanie tracheotomii w celu odsysania wydzieliny z dróg oddechowych. Pacjentka wymagała odsysania ssakiem nadmiernie gromadzącej się wydzieliny w jamie ustnej. Chorą oklepywano. Skóra pacjentki bez zmian patologicznych. Pacjentka z założoną gastrostomią. Podczas nocy pacjentka wymaga nieinwazyjnej wentylacji mechanicznej – respirator BIPAP. Pacjentka pod opieką Poradni Medycyny Paliatywnej. Pacjentka żywiona dietą przemysłową (Isosource Protein) do PEG – samodzielna w zakresie samoobsługi.

Wyniki. Pomimo, że choroba jest nieuleczalna, wiele jej objawów można złagodzić, a celem terapii powinna być poprawa jakości życia i podtrzymywania niezależności pacjenta tak długo, jak to możliwe. Pacjenci z SLA i ich krewni często doświadczają depresji, uczucia rozpaczy i lęku. Dlatego zalecana jest pomoc psychologiczna dla pacjentów i ich krewnych. Ciągły rozwój medycyny pozostaje nadzieją dla chorujących. Istnieje potrzeba kontynuacji badań naukowych w kierunku poszukiwania skutecznego leku na stwardnienie zanikowe boczne.

Abstract

Background. Amyotrophic lateral sclerosis is a degenerative disease of the nervous system, neurodegenerative cerebral cortex, pad and spinal cord due to damage to the upper and lower motor neurons, with progressive course, various symptoms with varying intensity and duration (from several months to even 10 years), extremely unfavorable prognosis. As an incurable and progressive disease, due to the deterioration of mobility it leads to complete stopping of muscles, including respiratory ones, with the intellectual ability of the patients fully preserved

Case report. A 65-year-old patient was hospitalized in Geriatrics Clinic in University Hospital A. Jurasza in Bydgoszcz. Patient with shortness of breath, present paradoxical chest movement, impaired chest mobility, present whirring, wheezing during auscultation examination. The patient was admitted to the ward because of pneumonia. Amyotrophic lateral sclerosis, chronic respiratory failure, hypertension, and hyperthyroidism were diagnosed. In the medical history she had swallowing problems, abdominal pain, insufficient cough reflex, dysphagia, increased body temperature, difficulty falling asleep, depressed mood. The patient did not consent to a tracheotomy for aspiration of secretions from the respiratory tract. The patient required suctioning of excessively accumulating secretions in the mouth. The patient was patted. Patient's skin without pathological changes. During the night, the patient requires non-invasive mechanical ventilation - BIPAP respirator. Patient under the care of the Palliative Medicine Clinic. Patient fed an industrial diet (Isosource Protein) for PEG (percutaneous endoscopic gastrostomy) - self-service.

Results. Although the disease is incurable, many of its symptoms can be alleviated, and the goal of therapy should be to improve the quality of life and maintain patient independence for as long as possible. SLA patients and their relatives often experience depression, feelings of despair and anxiety. Therefore, psychological help is recommended for patients and their relatives. The continuous development of medicine remains a hope for this disease. There is a need to continue research into the search for an effective cure for amyotrophic lateral sclerosis.

Background. The etiology of SLA (amyotrophic lateral sclerosis - SLA) is unknown. There are genetic risk factors for this disease. Recent studies on rare genetic forms of motor neuron disease point to the unique sensitivity of motor cells to glutamate excitotoxicity and disturbances in axonal transport, mitochondrial function and oxidative stress [1,3,4,12]. A direct relationship between a single environmental factor and the risk of SLA cannot be demonstrated. Probably only nicotinism can affect SLA risk, while the relationship to other risk factors is unclear. Most authors support the hypothesis of the complex etiology for SLA, which arises as a result of the interaction of genetic factors with environmental factors. The exact molecular mechanisms that cause motor cell degeneration are unknown [1,17]. Pathogenesis is also unknown. Genetic factors, excitotoxicity, oxidative stress, mitochondrial disorders, axonal transport disorders, aggregation of neurofilaments, inflammatory theory can have an impact on the emergence of the disease [5]. Deficiency of neurotrophic factors and dysfunction of cell signal transduction, histopathological features [3.8]. Amyotrophic lateral sclerosis is included in the group of motor neuron disease - according to WHO ICD-10 classification group G12.2. It is a degenerative disease of the nervous system, neurodegenerative cerebral cortex, bulb and spinal cord due to damage to the upper (UMN) and lower (LMN) motor neurons, with a progressive course, various symptoms with varying intensity and duration (from several months to even 10 years), with extremely unfavorable prognosis [1,10]. As an incurable and progressive disease, as a result of deterioration of mobility it leads to complete stopping of muscles, including respiratory ones, with the intellectual ability of patients fully preserved [16]. In the bulbar form, symptoms include dysarthria, dysphagia, muscular atrophy, fasciculation [2,10]. The patient feels emotional lability, exorbitant reflexes occur, e.g. mandibular or vivid throat reflex with simultaneous

paresis of speech organs [16,17]. Muscle weakness and atrophy, paddles and respiratory muscles occur. In the advanced stage, anartria occurs [2]. The peak incidence of SLA is observed in people aged 65 years. Within 2 years of diagnosis, 25% of patients die, half survive 3-4 years, although sporadic 5-10-year survival occurs (in over 20%). The patient's life expectancy depends primarily on the order in which the bulb and limb symptoms appear. The shortest survival time occurs when bulbar symptoms predominate and range from several months to 2 years. SLA is a rare disease. Its incidence is 1-2 / 100,000, and the incidence is 4-6 / 100,000. people. Death occurs mainly as a result of respiratory failure, pulmonary embolism due to patient immobilization or respiratory infection, most often in sleep [1.15]. Diagnosis of the disease: electrophysiological examinations, electroneurography studies (ENG), electromyography studies (EMG), transcranial magnetic stimulation (TMS), MUNE examination, magnetic resonance imaging (MRI) is used to detect changes in the corticospinal tract. Treatment of the disease: Despite many clinical trials and progress in understanding SLA pathologies, there are no drugs that effectively inhibit disease progression. Riluzol is the only drug with proven low efficacy in prolonging life in patients with SLA, patients often use replacement ventilation. An effective drug is being sought [3]. Stem cells are a relatively new and promising approach to SLA treatment [13]. The following drugs are under investigation: perampanel, tirasemtiv, pimozid, ranolazine, triheptanoin, edaravone, taurodeoxycholic acid [6,7,9]. Clinical trials have shown the beneficial effect of moderate exercise on the state of ALS patients. It is recommended to exercise: stretching, aerobic, resistance, strengthening and balance, as well as special methods such as the method Vojta and PNF. Literature data show that exercise influence on the activity of motoneurons which are more resistant to apoptosis selectively [14].

Case report. A 65-year-old patient was hospitalized in the Geriatrics Clinic in University Hospital A. Jurasza in Bydgoszcz because of pneumonia. A patient walking alone. Logical contact kept by gestures, facial expressions, nodding, writing by the patient on the tablet with the stylus. Patient with shortness of breath, present paradoxical chest movement, impaired chest mobility, present whirring, wheezing during auscultation examination. Amyotrophic lateral sclerosis, chronic respiratory failure, hypertension, and hyperthyroidism were diagnosed. In the history of swallowing problems, abdominal pain, insufficient cough reflex,

dysphagia, increased body temperature, difficulty falling asleep, depressed mood. The patient did not consent to a tracheotomy for aspiration of secretions from the respiratory tract. The patient required suctioning of excessively accumulating secretions in the mouth. The patient was patted. Patient's skin without pathological changes. During the night, the patient requires non-invasive mechanical ventilation - BIPAP respirator. Patient under the care of the Palliative Medicine Clinic. Patient fed an industrial diet (Isosource Protein) for PEG - self-service. Accepted medications for PEG after breaking down, administered together with water: riluzole PMCS 50mg, co-valsacor 80mg + 12.5 mg, euthyrox 100mg, sertagen 50mg, tussicom 200mg, amitriptylinum, paracetamol 500mg, Ceftazidime 1000 mg, fenoterol Ipratropium bromidum. The patient was given intermittent oxygen therapy at a flow rate of 1l / min. Basic parameters of the patient: Pressure 115/80; Pulse: 105; Temperature 38 Celsius degrees, BMI: 20.5. The results of the scales: ADL (Activities of Daily Living) - 5 points, IADL (Instrumental Activities of Daily Living) - 17 points, NRS (Numerical Rating Scale) - 3 points. In laboratory tests, an increase in indicators such as: white blood cells, platelets, red blood cell distribution width, platelet hematocrit, monocytes, C-reactive protein, glycemia, carboxyhemoglobin ; while a reduced indicator: red blood cells, hemoglobin, hematocrit, lymphocytes, eosinophils, potassium, serum creatinine, partial pressure of carbon dioxide, oxygen partial pressure, concentration total hemoglobin, deficiency of blood bases, oxygen saturation of hemoglobin, folic acid. Corynebacterium spp. was grown in microbiological tests.

Disscusion. An attempt to understand the essence of SLA is the international research program of scientists from the University of Sydney called ALS Quest. The essence of this commitment does not include the analysis of existing data, but the creation of a new set. The key to success is to obtain data from the widest possible population. Therefore, the tool - a questionnaire available for self-completion on the Internet - has been translated into 27 languages (among them Persian or Vietnamese as well as Polish) and this collection is constantly being expanded. The detailed survey contains a number of questions about the whole group of people - from general demography, through family life, professional life, habits, social interactions, psychology, hobby and environmental factors. The online questionnaire for the ALS Quest program was officially launched on January 30, 2015 [7].

Brain-computer interfaces use measurements of electrophysiological activity for extramuscular communication of man with the environment. In this way, the human central nervous system is able to connect to a computer that receives an EEG signal from the cerebral cortex. The connection with the human brain is invasive or non-invasive. The development of brain-computer interfaces can be associated with the robotization of workplaces, with the current state of research, only as one of the effects of technological development. It is possible, however, that this development will contribute to greater professional activation of people with disabilities, who through these interfaces will be able to work not only in the IT industries, but also as machine operators working in companies on an equal footing with non-disabled people. The brain-computer interfaces are constantly being improved because it is difficult to adapt the device to a human brain characterized by some variability. In some cases, the signals may be misinterpreted, or the device needs several attempts to read the information correctly [11]. The quality of life of patients with SLA would be better if professional institutional care was provided around the clock at home, which would allow other family members to function normally. This would allow the patient to be treated as a healthy family member, without a sense of responsibility in meeting the biopsychosocial needs of the sick person, which, as a result of long-term care, leads to exhaustion of caregivers and the patient's feeling that it is a burden on their surroundings, an obstacle to the implementation of other life plans of carers and others family members [1].

Conclusions. Although the disease is incurable, many of its symptoms can be alleviated, and the goal of therapy should be to improve the quality of life and maintain patient independence for as long as possible. SLA patients and their relatives often experience depression, feelings of despair and anxiety. Therefore, psychological help is recommended for patients and their relatives. The continuous development of medicine remains a hope for this disease. There is a need to continue research into the search for an effective cure for amyotrophic lateral sclerosis.

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