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Rare Diseases in Neurology — Caring for a Patient with Pompe's Disease

Choroby rzadkie w neurologii — opieka nad pacjentem z chorobą Pompego

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Abstract

Introduction. Pompe disease, a severe metabolic myopathy, is caused by mutations in the gene coding for acid alpha-glucosidase (GAA), what lead to intralysosomal accumulation of glycogen in all tissues, most notably in skeletal muscles. Pompe disease was the first documented lysosomal storage disease, nowadays we know around 60 similar disorders.
Aim. Presentation of the clinical picture of a man with Pompe's disease.
Case Report. A man at the age of 40, diagnosis of the Pompe's disease was made only at the age of 31. The first symptoms, indicating the patient's development of the disease, were already present in the early school age. At first, the clinical picture presented by the patient led to the diagnosis of muscular dystrophy.
Discussion. Pompe disease presents as a continuum of clinical phenotypes that differ by age of onset, severity, and organ involvement. Pompe disease affects people of all ages with varying degrees of severity. Two main broad types are recognized based on the onset of symptoms and the presence or absence of cardiomyopathy. Infantile onset Pompe disease (IOPD) as one, and the most severe for mod the disease. Other and less destructive is late-onset Pompe disease (LOPD) manifests any time after 12 months of age. The disease can be successfully treated by enzyme replacement therapy with alglucosidase alfa that was approved for human use in 2006.
Conclusions. In big importance is nurses role as educators and support for the patients during their hospitalizations for medicine infusions twice a month. It time when the knowledge and significance of proper life style can be discussed and implemented to empower the patients. **(JNNN 2019;8(4):170–176)**
Key Words: Pompe's disease, treatment, diagnosis, care

Streszczenie

Wstęp. Choroba Pompego, ciężka miopatia metaboliczna, jest spowodowana mutacjami w genie kodującym kwaśną alfa-glukozydazę (GAA), co prowadzi do wewnątrz lizosomalnej akumulacji glikogenu we wszystkich tkankach, zwłaszcza w mięśniach szkieletowych. Choroba Pompego była pierwszą udokumentowaną lizosomalną chorobą spichrzeniową, obecnie znamy około 60 podobnych zaburzeń.
Cel. Prezentacja obrazu klinicznego mężczyzny z chorobą Pompego.
Opis przypadku. Mężczyzna w wieku 40 lat ze zdiagnozowaną chorobą Pompego w wieku 31 lat. Pierwsze objawy wskazujące na rozwój choroby u pacjenta występowały już we wczesnym wieku szkolnym. Na początku obraz kliniczny przedstawiony przez pacjenta doprowadził do rozpoznania dystrofii mięśniowej.
Dyskusja. Choroba Pompego stanowi ciągłość klinicznych fenotypów, które różnią się w zależności od wieku pojawienia się objawów, jej ciężkości oraz dotkniętych narządów. Choroba Pompego w różnym stopniu nasilenia dotyka ludzi w każdym wieku. Dwa główne typy są rozpoznawane na podstawie pojawienia się symptomów i obecności lub braku kardiomiopatii. Choroba Pompego wieku dziecięcego (IOPD) stanowi jedną z najcięższych postaci choroby. Inne, mniej destrukcyjne odmiany choroby Pompego z późnym wystąpieniem symptomów (LOPD) pojawiają się w dowolnym momencie po 12 miesiącu życia. Chorobę można skutecznie leczyć za pomocą enzymatycznej terapii zastępczej alglukozydazą alfa, która została zatwierdzona do stosowania u ludzi w 2006 r.

Wnioski. Duże znaczenie odgrywa rola pielęgniarek, jako nauczycieli i wsparcie pacjentów podczas ich hospitalizacji wynikającej z infuzji leku dwa razy w miesiącu. Nadszedł czas, kiedy wiedza i znaczenie właściwego stylu życia mogą zostać omówione i wdrożone w celu wzmocnienia pozycji pacjentów. **(PNN 2019;8(4):170–176)**
Słowa kluczowe: choroba Pompego, leczenie, diagnoza, opieka

Introduction

Currently, 6% of the European society, or over 30 million people, suffer from rare diseases. Pompe's disease is one of almost six thousand known rare diseases. The essence of the disease is the mutation of the gene responsible for the breakdown of glycogen into glucose. Undecomposed glycogen is excessively stored in cells, mainly skeletal muscle and heart muscle, leading to significant impairment of their function.

Not much is said about rare diseases, which is why they are called orphan diseases in some regions of the world. These diseases are associated with persistent treatment that is difficult to access. Patients with Pompe's disease in Poland have been able to benefit from free treatment for 12 years, which consist in supplying/replacing the missing enzyme responsible for glycogen breakdown [1].

Patients with Pompe's disease who have the opportunity to receive treatment to replace the missing enzyme are subjected to daily hospitalization, during which check-ups, assessment of the patient's health and intravenous administration of the drug are performed. The drug infusion is administered by a pump, under the supervision of a neurologist and nursing staff of neurological departments.

The aim of this work is presentation of the clinical picture of a man with Pompe's disease.

Pathogenesis of the Pompe's Disease

Pompe's disease is a rare genetic disease inherited in an autosomal recessive manner. It is also called the type II glycogenosis. It was first described in 1932 by the Dutch pathologist, Johannes C. Pompe. The essence of the disease is the defect in the GAA gene — coding the lysosomal enzyme α-1-4-glucosidase, which is responsible for the process of glycogen breakdown into glucose. As a consequence of this defect, the polysaccharide accumulates in the body's cells [2–4]. The most common location of glycogen deposits is skeletal muscle and heart muscle, but they also accumulate in smooth muscles and in glial cells, brain stem nuclei and spinal cord, preventing them from working properly.

The first emerging dysfunctions resulting from type II glycogenosis are caused, as is well known, by intra-lysosomal glycogen accumulation. However, studies have shown that there are many other cellular abnormalities making the pathophysiology of Pompe's disease much more complex than it was previously thought, which is mainly associated with cellular autophagy disorders. As a result, the abnormalities in the genetic record, the GGA work may be impaired, have an abnormal course or not occur at all due to excess glycogen accumulation in the lysosome [5]. Pompe's disease approximately occurs once in every 40.000 births. It affects less than 200.000 people in the United States and no more than 5.000–10.000 in Europe (1/283.000). People of all races can get glycogenosis, but African Americans have it more often (1/14.000 births) than white people (1/60.000 adults and 1/140.000 children). Due to unspecific symptoms and rarity, thousands of patients with unclassified musculoskeletal symptoms may have the undiagnosed Pompe's disease. In many cases, the final diagnosis falls even 8 years after the appearance of the first symptoms of the disease [6,7].

The clinical manifestations of the Pompe's disease show a wide variety. They depend mainly on the degree of the retained enzyme activity. The complete inactivity is associated with early and severe symptoms.

The Clinical Image of the Pompe's Disease

Depending on when the first symptoms appear, we distinguish the childhood, adolescent and adulthood form of the Pompe's disease. Both the age at which the person's symptoms appear and the degree of organ involvement have a lot in common with GAA enzyme activity: in infants, it is less than 1% of normal activity, in adolescents less than 10%, in adults less than 40% [8].

Infant Form of the Pompe's Disease

The most severe form of the Pompe's disease occurs in infancy. Symptoms often affect several systems at once (Table 1) [8,9].

Table 1. Symptoms of the infant form of the Pompe's disease

Gastrointestinal symptoms:	Cardiovascular symptoms:
— difficulty in taking food,	— congestive heart failure,
— organomegaly (among others, hepatomegaly, tongue hypertrophy).	— hypertrophic cardiomyopathy.

Table 1. Continued

Respiratory symptoms: — frequent respiratory infections, — aspiration pneumonia, — breathing disorders during sleep, — progressive respiratory failure.	Musculoskeletal system symptoms: — reduced muscle tone, — muscular weakness, — drooping head.

Adolescent Form of the Pompe's Disease

In the adolescent form of the Pompe's disease, the flagship symptom is the progressive weakens of the proximal limb muscles. It is also characteristic to delay motor development resulting in difficulties in moving upright. There are also difficulties in swallowing food and breathing disorders due to damage to the respiratory muscles by a progressive disease process. Due to the accumulation of glycogen in the muscles, they become hard and change their consistency. Calf muscle hypertrophy and the emerging Gowers symptom (climbing on itself) often suggest Duchenne muscular dystrophy. However, these symptoms occur at a younger age than with dystrophinopathy. Pompego cardiomyopathy occurring in the adolescent form is usually mild and does not affect the obstruction of the outflow tract from the left ventricle [9].

Among patients with the Pompe's disease in the adolescent form, death usually occurs before the end of the second decade of life.

Form of the Pompe's Disease Occurring in Adults

In the late-onset Pompe's disease, proximal paresis is the main symptom. Patients then struggle to get up the stairs, raise their arms or get out of the chair. Table 2 presents symptoms related to other systems [9].

The picture of symptoms in the adult form of the Pompe's disease is associated with the girdle-muscular dystrophy. The coexisting respiratory failure develops slowly and chronically, manifesting itself in shortness of breath after exercise, which makes patients sometimes be unaware of it. If you undergo surgery under general anaesthesia, there may be a significant reduction in your breathing capacity. Unlike the forms of the Pompe's disease described above, myocardial symptoms are very rare in adults. In many cases, patients who fell ill as adults had previously had normal life activities and did not notice any limitations in their physical capacity [9].

Diagnosis and Additional Tests Supporting the Diagnosis

The definitive diagnosis of the Pompe's disease is difficult for diagnosticians because its symptoms are similar to many other, more common disorders. This often affects significant diagnostic delays in many patients. Diagnostic tests performed for Pompe's disease include, among others: skin or muscle biopsy, laboratory blood tests, electromyography, echocardiography, dry blood test or genetic tests [5].

Skeletal muscle biopsy is one of the most significant diagnostic methods for neuromuscular diseases. It allows detection of many specific changes in muscle structure, such as membraned glycogen in Pompe's disease. Comparing the results of histopathological tests with the results of electrophysiological tests largely gives the possibility of making a diagnosis. The additional use of immunohistochemical techniques allows for closer characterization, for example, glycogenesis. The muscle that will be selected for biopsy should show clinical signs of the disease. The reduced level of lack of α-1-4-glucosidase activity and the presence of vacuoles in the biopsy from the diseased site, which are staining positively towards the presence of glycogen, allows fora reliable diagnosis. Due to the availability of much less invasive

Table 2. Symptoms of the Pompe's disease in the adult form

Gastrointestinal symptoms: — problems with swallowing, chewing, tiredness of the jaw muscles, — low weight gain and difficulty with weight maintenance, — elevated liver enzymes concentration.	Respiratory symptoms: — shortness of breath which worsens in the supine position, — exertional dyspnoea, — respiratory infections, — sleep apnoea, night hypoventilation, — daytime sleepiness, — morning headaches.
Musculoskeletal system symptoms: — muscle weakness, in particular near the torso, — posture disorders, — difficulty climbing stairs, — frequent falls and muscle pains.	

tests, muscle biopsy has relatively less diagnostic significance. However, it can be valuable in cases of doubt among doctors. About 20–30% of patients with the form of the disease occurring in adulthood do not have typical ultrastructural changes [10].

Concentrations of creatine kinase in the serum, aspartate transaminase, lactate dehydrogenase and myoglobin are determined in laboratory tests to diagnose Pompe's disease. Enzyme activity of α-1-4-glucosidase can also be determined in peripheral blood leukocytes.

Electromyography is used in neuronal muscle disorders. This test involves placing electrodes on the skin to monitor the activity of individual muscles. In the case of Pompe's disease, electromyographic test often shows the features of myopathy with hyperactivity of muscle fibres and pseudotonic discharges [11].

Myocardial echocardiography shows its large hypertrophy in new born babies and infants with early form of the disease. In the adolescent and adult forms, heart enlargement is much milder or absent.

The dry blood test is an enzymatic screening test for Pompe's disease. It should be carried out in every case of unexplained muscle symptoms after excluding the most common causes of myopathy. Blood is placed in the paper and sent to the laboratory to determine the α-1-4-glucosidase activity. In Poland, dry blood screening tests are currently free of charge [11]. In some countries, a new born screening program for Pompe's disease has been introduced using this diagnostic method. These were the United States (Washington, Missouri, Illinois), Taiwan, Japan, Italy, Germany, Columbia and Austria. Despite the acceptance of screening tests for lysosomal storage diseases (including Pompe's disease) in medical environments, some researchers and ethicists recommend for them to be carried out only after prior approval by the scientific committee and with the consent of the parents [12].

Significant reduction of α-1-4-glucosidase activity or lack thereof in the enzymatic test requires confirmation by genetic testing. It should show mutations in both alleles of the GAA gene. In patients with an infant form, both alleles have mutations that cause almost complete inactivation of the gene. In late-onset disease, at least one of the alleles has a mutation that produces a weaker pathogenic effect.

Additional Tests

In diagnosed infants, Western blot testing for the presence of α-1-4-glucosidase in skin fibroblasts may be indicated prior to initiating enzyme replacement therapy. This allows the detection of specific enzyme antigens. In the complete absence of α-1-4-glucosidase, the patient's body may not be able to develop immune tolerance, as a result of which inactivating antibodies may appear after administration of the exogenous enzyme drug, which limits the effectiveness of the treatment. Spirometry, which should be performed in every cooperating patient with myopathy, can confirm the impairment of respiratory function [13].

The Treatment of Pompe's Disease

The treatment of Pompe's disease involves the implementation of therapy specific for the patomechanism of the disease and parallel supportive treatment. Patients affected by the disease have a wide range of clinical symptoms associated with functional impairment from various systems. Therefore, it is important that the therapeutic process is led by a multidisciplinary team led by an experienced neurologist, which includes a cardiologist, pulmonologist, orthopaedist, physiotherapist and dietician [2].

In the past, treatment for Pompe's disease has been mainly focused on symptom relief and supportive care. The standards of care published by the American College of Medical Genetics (ACMG) currently direct the procedure towards quick diagnosis and personalized causal treatment.

Undoubtedly, the most important role is attributed Enzyme Replacement Therapy — ERT. In 2006, the Food and Drug Agency approved and permitted this medical product as the first and only one that can be used in this type of treatment for Pompe's disease. Alglucosidase alfa restores the activity of lysosomal acid glucosidase alpha, leading to the stabilization or reconstruction of the functions of the heart muscle, skeletal or respiratory muscles. The human acid α-1-4-glucosidase contained in the drug is obtained by recombinant DNA method in Chinese hamster ovary cells. The recommended dose is 20 mg/kg of body weight and is administered once every two weeks. The response to treatment should be regularly evaluated, carefully analysing all clinical manifestations of the disease [1].

The indication for the administration of the drug includes patients of all ages. Therapy should be conducted by a physician experienced in the treatment of patients with metabolic or neuromuscular diseases. The treatment product should be administered as an intravenous infusion under hospitalization conditions. In Great Britain and the Netherlands, infusions are also performed at home [7].

In the case of a milder form of the disease, when there is no need for enzyme replacement therapy, it is important to include a diet low in carbohydrates and rich in protein [14]. Increased supply of amino acids, which are a substrate in protein synthesis, may weaken muscle tissue proteolysis. The amount of carbohydrates

consumed during the day should not only be reduced to 30–35%, but also distributed over time. Applying the "little and often" principle helps to avoid glycogen build-up and at the same time prevents hypoglycaemia. Recommended products are presented in Figure [14,15].

In advanced cases of Pompe's disease, when patients develop respiratory failure or sleep apnoea syndrome, non-invasive mechanical ventilation in required as part of the treatment. Movement improvement is also very important in the treatment of all forms of the disease.

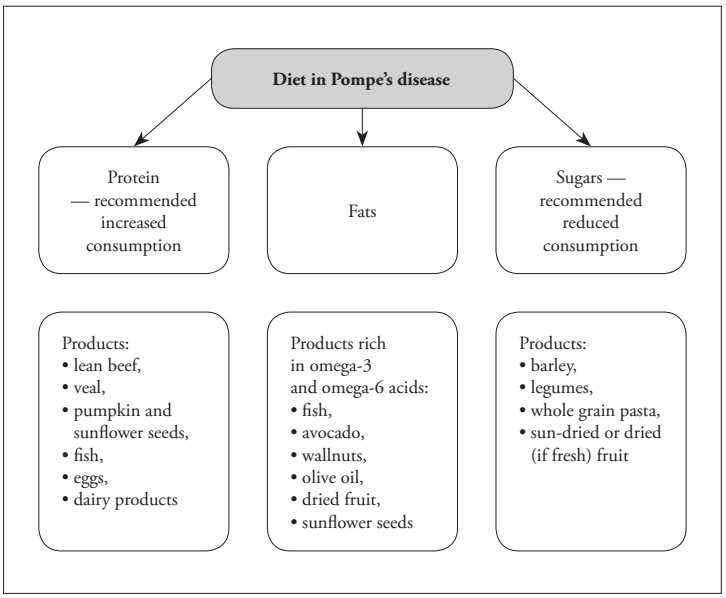

Figure. Dietary recommendations in Pompe's disease [14]

Case Study — Caring for a Patient with Pompe's Disease

A man at the age of 40, diagnosis of the Pompe's disease was made only at the age of 31. The first symptoms, indicating the patient's development of the disease, were already present in the early school age. In an interview, the patient reported that since he can remember, he had a big problem with physical exercises, which was manifested especially during physical education classes. The patient's performance was much weaker than that of his peers. Symptoms of weakness and fatigue were associated with the prematurity of both the discussed patient, and his younger sister, who also took her first steps very late (also with a diagnosed Pompe's disease).

At first, the clinical picture presented by the patient led to the diagnosis of muscular dystrophy. In 2011, after cementoplasty under general anaesthesia, there was a significant deterioration in the patient's respiratory capacity and the need for respiratory therapy. In-depth diagnostics, carried out due to numerous fractures and deterioration of the patient's condition, including electromyographic examination and specialized neurological consultations in Warsaw, led to registration in a genetic outpatient clinic. In 2011, Pompe's disease was diagnosed with a dry blood test in both the patient and his sister. For a year, the patient and the therapeutic team applied for inclusion in the National Health Fund program called "Treatment of Pompe's Disease", which provides reimbursement of the alglucosidase alfa drug. Since September 2012, the patient has been qualified and receives an intravenous infusion every two weeks enabling the further functioning. Currently, the patient has muscle atrophy, muscle pain, gait disturbance. To move, the patient requires auxiliary equipment, he uses an elbow orthopaedic crutch. Past and coexisting diseases: hypertension, respiratory failure requiring CPAP respiratory therapy, condition after vertebral Th12, L1, L3, L4, L5 fracture and cementoplasty of the L1 and L5 vertebra (2011 and 2012), diverticulitis, left subtrochanteric femur fracture. Previous surgeries: cementoplasty of L1 and L5 (2011 and 2012). In 2016, open reposition and DCS anastomosis of the left femoral subtrochanteric fracture, in 2017 removal of the fusing material, complicated by cardiac arrest.

Assessment of the Patient's Condition

Respiratory system:
— weak breathing, shallow breathing, through an abdominal track, regular 16/min, through the nose;
— shortness of breath;
— weakness of the breathing muscles;
— home respiratory therapy — CPAP, used at night and when resting in the supine position.

Cardiovascular system:
— correct blood pressure 116/80 mmHg, the patient takes hypertension medicines;
— heart rate 60 beats/min, well felt on the radial artery.

Digestive system:
— diverticulum;
— periodic stool incontinence;
— normal BMI (24.5 kg/m^2);

Genitourinary system:
— efficient diuresis, without a diuretic;
— the patient uses the toilet by himself;
— sight: no significant deviations;
— hearing: no significant deviations;
— smell: no significant deviations;
— taste: no significant deviations.

Nervous system:
— the patient oriented in terms of place and time;
— verbal and logical contact preserved;

— euthymia;
— correct circadian rhythm.

Osteoarticular system:
— weakness of the abdominal muscles, spine, hips and lower limbs;
— condition after fractures of the vertebra Th12, L1, L3, L4, L5, and cementoplasty of L1 and L5;
— the patient uses the elbow orthopaedic crutch just in case.

Skin:
— clean;
— no pressure sores (17 points on the NORTON scale);
— no breach of continuity.

Results of Blood Tests Performed on the Day of Admission

Blood morphology:
— Haemoglobin: 13.8 g/dl (14–18 g/dl),
— Haematocrit: 43.2% (40–54%),
— Red blood cells: 4.74 T/l (4.5–5.4T/l),
— Platelets: 307 G/l (150–400 G/l),
— Leukocytes: 9.23 T/l (4–10 T/l).

Acid-base balance:
— pH: 7.37 (7.32–7.42),
— pCO_2: 47.8 mmHg (35–45 mmHg),
— pO_2: 61.4 mmHg (25–40 mmHg).

Biochemistry:
— Sodium: 140 mmol/l (135–145 mmol/l),
— Potassium 4.7 mmol/l (3.5–5.2 mmol/l),
— Creatine kinase: 445 U/l (55–370 U/l).

Hepatic enzymes:
— Aspartate aminotransferase: 46 U/l(<40IU/l),
— Alanine aminotransferase: 44 U/l(<40IU/l).

Electrocardiogram: sinus rhythm, steady heart rate, without signs of recent myocardial ischemia.

Support in the Pompe's Disease

Due to its progressive nature and specific form of treatment, support has a very large impact on the quality of life of patients and their families or carers. It is noted that the patient's mental state is closely related to the extent of clinical manifestations of the disease. Undoubtedly, this is associated with muscle pain, physical weakness, difficult breathing or limitation in everyday activities. The need to undergo hospitalization twice a month, change in lifestyle or struggle with serious symptoms makes a patient with Pompe's disease a challenge for a nurse related to mental and spiritual care [16–18].

Showing empathy in conversation with a patient, the ability to listen actively and show him due attention can reduce the level of stress and anxiety associated with subsequent hospitalization. It is important to use a large number of positive messages and constantly make the patient aware of his strengths. Each conversation should take place in accordance with intimacy conditions and at a jointly scheduled time. In many patients affected by genetic diseases, improving mental state is influenced by allowing contact with another person struggling with a similar or the same disease. It is also helpful to provide the patient with information brochures, phone numbers or website addresses for associations of people with type II glycogenosis. In cases of significantly reduced well-being, the patient may be advised to seek professional psychological support [16,18].

Psychological care should also often include caregivers and people close to the patients. Caring for a disabled person with a life-long disease perspective is not only a physical burden for them, but also a mental one. Family members devoted to caring for a sick person are often forced to give up their previous lives. Therefore, their existential needs should also be taken into account in diagnosing biopsychological problems as part of the nursing process [16,18–20].

Conclusions

Current management of Pompe disease is focused mainly on the enzyme replacement treatment but we should not forget about the importance of the diet and exercise as the support for the general condition of the patients. In big importance is nurses role as educators and support for the patients during their hospitalizations for medicine infusions twice a month. It time when the knowledge and significance of proper life style can be discussed and implemented to empower the patients.

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