The Patient's Way to Diagnosis: A Clinical Case of Late Onset Pompe Disease in an Adult

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ABSTRACT

BACKGROUND: *Late onset Pompe disease is a rare genetic disease from the group of the accumulation diseases, the main manifestation of which is the progressive myopathic syndrome. The difficulties of diagnostics are mainly related to the low awareness of the specialized physicians (neurologists, orthopaedists, rheumatologists, pulmonologists, pediatricians etc.) on the specific features of this disease, as well as to the low level of nosological specificity of the myopathic syndrome in general. Special importance of early diagnostics is due to the existence of pathogenetic therapy. Late diagnostics and delayed initiation of therapy lead to lower survival rate and higher rate of incapacitation among the patients.* ***CLINICAL CASE DESCRIPTION:*** *The patient aged 62 years, which for many years was under the supervision by neurologists with the diagnosis of osteochondrosis, based on the objective data, actually had a myopathic syndrome that was diagnosed and confirmed using the electroneuromyography. The detected findings included a decrease in the activity of the alpha-glucosidase enzyme, while the genetic examination that followed, allowed for detecting the presence of a mutation in the GAA gene. The dynamic changes of the disease were tracked with a background of taking pathogenetic therapy for 4 years.* ***CONCLUSION:*** *This clinical case demonstrates the many years of the patient's way to being diagnosed with a rare genetic disease and, respectively, to the later initiation of therapy. The efficiency of treating the accumulation diseases directly depends on the extent of the pathological changes in the target organs, accumulated to the moment of diagnostics, which means — the earlier, the more effective.*

Keywords: *late onset Pompe disease; myopathy; enzyme replacement therapy; glycogenosis type II; accumulation diseases.*

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BACKGROUND

Late onset Pompe disease is a rare disease, which relates to type II glycogenosis and is a hereditary accumulation disease with an autosomal-recessive mode of inheritance. The clinical manifestations of the disease are determined by the accumulation of glycogen in the lysosomes of the skeletal muscles, of the parenchymatous organs, of the nervous system and of other tissues. The accumulation of glycogen is caused by the insufficiency of the enzyme — the acid alpha-glucosidase. The insufficiency of the enzyme develops as a result of *GAA* gene mutation (glucosidase alpha acid), localized at a long arm of the 17th chromosome.

The incidence of the disease in various countries ranges from 1:300 000 to 1:40 000 [1–3]. In Russia, the Pompe disease is included into the list of orphan diseases, but its incidence is unknown. As of the year 2024, in Russia this diagnosis was set to approximately 100 patients[1].

The majority of patients go through a very long way to setting the diagnosis and receiving the pathogenetic therapy. The low level of diagnostics is also due to the insufficient awareness of the pediatricians and neurologists on the specific features of the disease. The knowledge about this disease must also be available to the physicians of other specialties: pulmonologists, orthopaedists, rheumatologists and internists [4]. The difficulties of diagnostics are primarily related to the high variability of the disease onset age. Late onset Pompe disease can manifest both at an early age

[1] RG.RU [Internet]. Nevinnaya I. The experts: we need faster solving for the issue of including the Pompe disease into the neonatal screening // *Russian newspaper*. April 15, 2024. Access mode: https://rg.ru/2024/04/15/eksperty-nuzhno-bystree-reshit-vopros-s-vkliucheniem-v-neonatalnyj-skrining-bolezni-pompe.html

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Путь пациента к диагнозу: клинический случай болезни Помпе с поздним началом у взрослого

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АННОТАЦИЯ

Обоснование. *Болезнь Помпе с поздним началом — редкое генетическое заболевание, относящееся к болезням накопления, основным проявлением которого является прогрессирующий миопатический синдром. Трудности диагностики связаны в основном с низкой осведомлённостью врачей-специалистов (неврологов, ортопедов, ревматологов, пульмонологов, педиатров и др.) об особенностях этого заболевания, а также с низким уровнем нозологической специфичности миопатического синдрома в целом. Особая важность ранней диагностики обусловлена существованием патогенетической терапии. Поздняя диагностика и отсроченное начало терапии ведут к меньшей выживаемости и большей частоте инвалидизации пациентов.* ***Описание клинического случая.*** *У пациентки 62 лет, много лет наблюдавшейся у неврологов с диагнозом остеохондроза, объективно был выявлен миопатический синдром, подтверждённый с помощью электронейромиографии. Обнаружено снижение активности фермента альфа-глюкозидазы, а последующее генетическое исследование позволило определить наличие мутации в гене GAA. Прослежена динамика заболевания на фоне приёма патогенетической терапии в течение 4 лет.* ***Заключение.*** *Данный клинический случай иллюстрирует многолетний путь пациентки с редким генетическим заболеванием к диагнозу и, соответственно, более позднему началу терапии.*

Ключевые слова: *болезнь Помпе с поздним началом; миопатия; ферментозаместительная терапия; гликогеноз II типа; болезни накопления.*

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(after 1 year of age) and at the older age. The main problem of the diagnostics is the non-specificity of clinical manifestations, the phenotype of patients with late onset Pompe disease is similar to a large number of neuro-muscular diseases. Due to the fact that late onset Pompe disease belongs to the group of myopathies, among the clinical signs, the ones prevailing are the muscle weakness and the respiratory abnormalities, which slowly progress and do not show clear nosological specificity. The diagnostics of Pompe disease is based on the screening of the activity of the acid alpha-glucosidase enzyme[2], which is supported by the molecular-genetic testing of the *GAA* gene upon detecting the decreased activity of the enzyme [5–7].

All the patients with Pompe disease should receive lifetime pathogenetic enzyme replacement therapy. Currently, in the Russian Federation, two medicinal products are registered: Alglucosidase alfa (from 2013) and Avalglucosidase alfa (from 2023). Avalglucosidase alfa, according to the data from clinical research, is a more effective medicinal product with better safety profile [8].

CLINICAL CASE DESCRIPTION

Patient info

Female patient X. born in 1959, office worker. Presented to the Neuromuscular Diseases Office of the Consultative and Diagnostic Center of the Federal State Budgetary Scientific Institution “Scientific Research Institute for Complex Problems of Hygiene and Occupational Diseases” (SRI CPH and OD) in March 2021 with the complaints of weakness in the limbs;

[2] Ministry of Health of the Russian Federation. List of headings of the clinical recommendations [Internet]. Pompe disease. The Union of Pediatricians of Russia, the Association of Medical Geneticists, Society of Specialists on Neuromuscular Diseases, 2019. Access mode: https://cr.minzdrav.gov.ru/view-cr/317_1

difficulties when walking up (the stairs, in the transport vehicles, walking uphill); when standing from the sitting position or lying position; changes in the gait; periodical spasms in the gastrocnemius muscles; shortness of breath when walking and upon mild physical activity.

Case history. The early development of the patient was unremarkable. From the school age, she had noted problems during physical exercises at school: difficulties were experienced when jumping or running (the only "C" mark during the school years was for physical education). From 2004/ 2005 (at the age of about 45 years), she started noting difficulties when standing up from the squat on haunches, when walking up the stairs of the vehicle; slight limping has developed. After 3 more years, a clear gait change was observed. During the last year, shortness of breath developed. The disease course is progressing. The relatives had no such symptoms. From the age of 30 years, the patient periodically undergoes check-ups at the neurologist office due to osteochondrosis with short-term effect.

Regular medical check-ups (follow-up). Disability group II (mastectomy in 2005 due to the malignant neoplasm, radiation and chemotherapy were used); hypothyroidism from 2006 (replacement therapy — receiving Euthyrox at a dosage of 100 μg).

The presence of injuries or infectious diseases — negative (verbal information provided by the patient).

Laboratory and instrumental diagnosis

Objective findings: clear consciousness, adequate behavior; oriented in the location, time and personality. The body constitution is correct, normosthenic. Height 172 cm, weight 82 kg. The skin surface is clean, showing usual coloring. Breath rate — 18 per minute, no rales were found in the lungs. Upon auscultation, the heart tones are rhythmic, the heart rate is 75 bpm, the blood pressure level is 120/78 mm.Hg. The abdomen is soft and painless. The functions of the pelvic organs are not impaired.

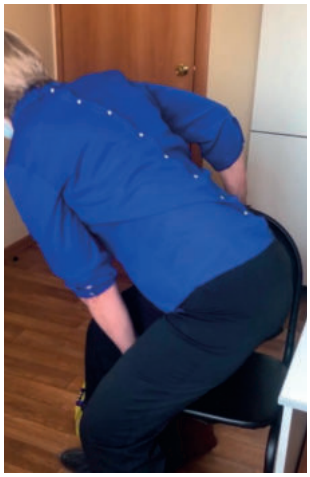

Fig. 1. Gower's maneuver upon standing.

Trendelenburg gait, the patient experiences significant difficulties when standing up from the sitting position, from the lying position, from squat on haunches, using the Gower's maneuvers (weakness of axial muscles) (Fig. 1, Supplement 1).

Craniocerebral nerves: the patient is capable of differentiating the smells, the fields of vision upon quick testing are normal, the palpebral fissures and the pupils are equal, active photoreactions are present, the ocular motility has a full range, no signs of nystagmus were found, the facial sensitivity is not impaired, the face is symmetrical, the phonation is intact, the throat reflexes show medium degree of activity, the tongue is positioned along the midline, no fasciculations were noted (visible involuntary contractions in separate bundles of muscle fibers).

Muscles. Mimic muscles — 5 points of strength according to the MRC scale (Medical Research Council). Strength in the proximal segments of the upper limbs: 4–5 points, in the extensor muscles of the forearms — 4 points, in the proximal segments of the lower limbs — 3–4 points; more decreased strength of the extensors and the adductors of the thighs, along with the extensor muscles of the shins. Visually, there are no signs of muscle atrophy or pterygoid scapulas.

Reflexes. The tone in upper and lower limbs is decreased, the tendon reflexes in the upper and lower limbs are decreased, no pathological reflexes were found. The abdominal reflexes are decreased (flaccid anterior wall of the abdomen), no sensitive disorders were observed. Romberg stance test — stable, coordination tests — satisfactorily.

The specific complaints and data from the neurological assessment allowed for suspecting the presence of primary muscular disease.

The provisional diagnosis set was the following: "Myopathic syndrome of unclear origin".

Clinical blood and urine tests were showing no abnormalities, except for periodical elevation of creatinphosphokinase (CPK) from 300 to 500–600 U/l.

Data from stimulation electroneuromyography conducted on the day of presenting to the Clinic of the SRI CPH and OD in March 2021: the conduction of impulse in the nerves of the upper and lower limbs is not impaired. Upon the needle electromyography of the muscles of the upper and lower limbs, single spontaneous activity was registered in the proximal muscles, the movement unit potentials in the distal muscles are intact, in the proximal muscles of the upper (the deltoid) and lower limbs (the quadriceps, the femoral adductor), the movement unit potentials have moderate remodeling of muscle type expressed as a decrease in the mean duration (Fig. 2).

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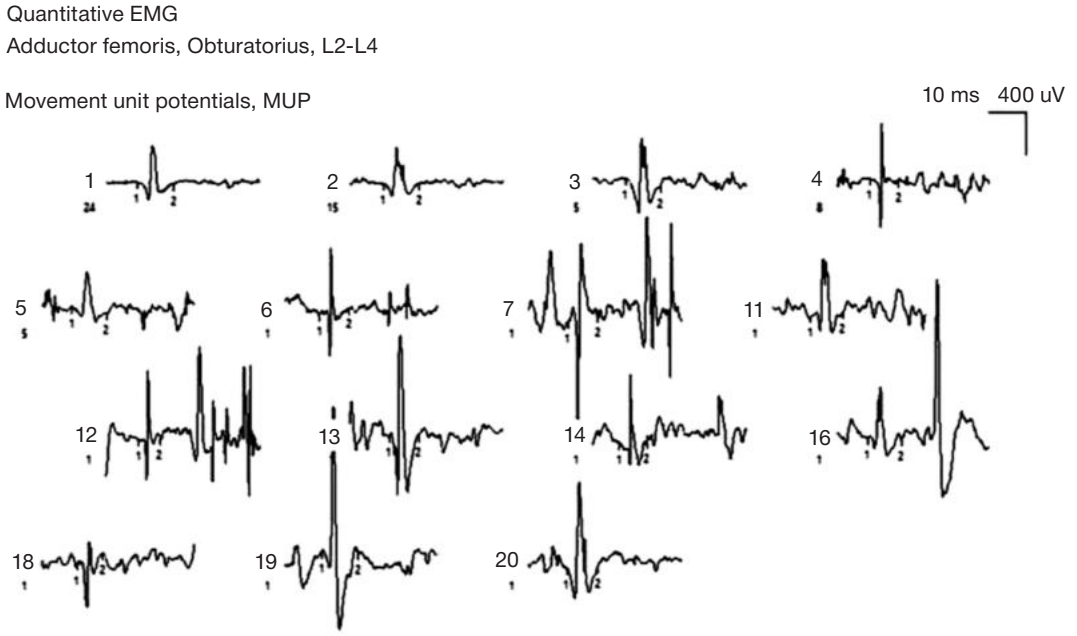
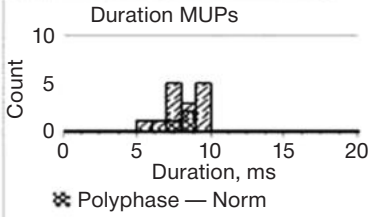


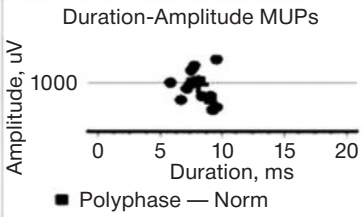
Duration MUPs

	Min duration, ms	Max duration, ms	Average duration, ms	Stage	Quantity MUPs
Total	5.8	9.48	8.08		15
N/p	5.8	9.48	8.05		12
P/p	7.52	8.98	8.19		3

Amplitude MUPs

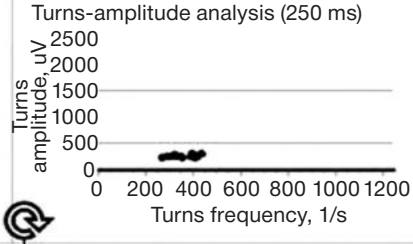
	Min amplitude, uV	Max amplitude, uV	Average amplitude, uV	Polyphase, %
Total	457	2057	940	20.0
N/p	457	2057	949	
P/p	639	1075	905	





Interpretation

Parameter	Malue
Fibrillation	Single
Positive sharp wave	No
Fasciculation	No
Amplitude MUPs	Normal
Duration MUPs	Significantly reduced
Polyphasy os MUPs	No
Type of interference pattern	Saturated reduced
Muscle pattern	Myogenic



Parameters MUPs

N	Take into account in the analysis	Duration, ms	Amplitude, uV	Area, nVxs	Thickness, ms	Phases	Turns	Risetime, ms	Periods per second	Peak duration, ms	Size Index
1	✓	9.46	496	788	1.59	3	3	0.775	8.1	2.68	0.979
2	✓	9.23	458	706	1.54	3	7	1.1	4.9	4.43	0.864
3	✓	9.02	689	1236	1.79	3	5	0.6	0.9	2.88	1.47
4	✓	7.32	957	445	0.465	3	5	0.3	2.0	3.93	0.427
5	✓	9.12	457	842	1.84	3	5	1.23	1.4	3.78	1.16
6	✓	7.52	1001	532	0.531	5	12	0.3		1.58	0.532
7	✓	7.65	1674	1654	0.988	3	7	0.75		5.0	1.44
11	✓	8.25	669	1059	1.58	4	8	0.65		3.13	1.23
12	✓	5.8	1022	456	0.447	4	8	0.275		1.6	0.466
13	✓	7.45	1463	2037	1.39	3	6	0.575		2.6	1.72
14	✓	7.13	842	672	0.797	3	9	0.2		2.35	0.648
16	✓	8.98	639	875	1.37	5	7	0.375		3.4	0.98
18	✓	6.65	603	566	0.94	4	10	0.35		1.65	0.5
19	✓	9.48	2057	2801	1.36	4	7	0.775		2.78	1.99
20	✓	8.07	1075	1567	1.46	5	5	1.0		3.73	1.52

Fig. 2. Myogenic remodeling of the movement unit potentials upon the needle electromyography (decreased duration, the saturated pattern). N/p — Non polyphase, P/p — Polyphase.

Molecular-genetic examination. In March 2021, blood sample was drawn for dried blood spot testing: the detected activity of the alpha-1.4-glucosidase was 0.36 μmol/l per hour (ref. ranges >2.32).

Using the method of next generation sequencing (NGS; NGS-panel), in the *GAA* gene, the pathogenic chr17:78078341T>G and chr17:78078692T>G nucleotide variants were detected in the heterozygous state. The examination was carried out at the Laboratory of Molecular Genetics and Medical Genomics of the Center for Fundamental Research in Pediatrics under the Federal State Autonomous Institution "National Medical Research Center for Children's Health", the subdivision of the Ministry of Health of the Russian Federation, the results were obtained in May 2021.

Definitive clinical diagnosis

Thus, the characteristic complaints, the anamnestic data, the data from neurological assessment, as well as the decreased activity of alpha-1.4-glucosidase in blood along with the confirmation of the presence of mutations in the *GAA* gene has allowed for stating the definitive clinical diagnosis: "Late onset Pompe disease".

Additional examination

The additional examination of the female patient was carried out at the Neurology Department of the Federal State Budgetary Scientific Institution "Scientific Center for Neurology" in June 2021, where the following results were obtained.

Laboratory tests. Alanine aminotransferase (GPT) 26 U/l, Aspartate aminotransferase (GOT) 59 U/l, Creatinphosphokinase (CPK) 516 U/l, Lactate dehydrogenase (LDH) 190 U/l, Creatinphosphokinase-MB (MB-CPK) 8 U/l (the reference ranges from the laboratory were not listed in the discharge epicrisis).

Instrumental examinations:

- echocardiography: the linear dimensions of the heart and the hemodynamic parameters in the heart valves are matching the age-specific reference ranges. The local and the global systolic function of the left ventricle is not impaired. Type I diastolic dysfunction of the left ventricle (impaired relaxation). Mitral regurgitation grade I. Tricuspid regurgitation grade I. No signs of pulmonary hypertension were detected;
- evaluation of the pulmonary functions test findings: vital capacity (VC) at the sitting position is 2.2 L (64.9% of the normal VC);
- magnetic resonance imaging (MRI) of the muscles: the data correspond to atrophy in thigh muscles with the presence of zones of swelling (stage 2b acc. to the classification by E. Mercuri, 2002).

Treatment

The enzyme replacement therapy with alpha-glucosidase medicinal product was prescribed to the patient. For two years (in 2021–2022) she was receiving Alglucosidase alfa, while the last two years (2023–2024) she was switched to Avalglucosidase alfa.

With the background of the pathogenetic therapy, the female patient subjectively reports positive dynamic changes expressed as an increase in the tolerance to physical activity along with decreasing shortness of breath.

Additional examinations conducted for dynamic follow-up at the Neurology Department of the Federal State Budgetary Scientific Institution "Scientific Center for Neurology" in August 2023, have demonstrated the following results:

1) laboratory tests: GPT 36 U/l, GOT 55 U/l, CPK 293 U/l, MB-CPK 30 U/l, LDH 162 U/l, alkaline phosphatase 189 U/l;
2) Instrumental examinations:
   - needle electromyography: the amplitude and the duration of movement unit potentials in the *musculus deltoideus* on the right side, in the *musculus biceps brachii* on the left side, in the *musculus paravertebralis* (Th12) on the left side and in the *musculus tibialis anterior* on the left side are within the normal ranges; in the *musculus biceps brachii* on the left side, there are single myogenic movement unit potentials; the amplitude of the movement unit potentials in the *musculus interosseus dorsalis* on the left side and in the *musculus vastus lateralis* on the right side is increased, the duration is within the normal ranges; other registered findings include spontaneous activity expressed as positive sharp waves in the *musculus interosseus dorsalis* and in the *musculus tibialis anterior* on the left side;
   - pulmonary functions test: VC 2.530 l (75.3% of the proper VC) at the sitting position;
   - echocardiography: signs of connective-tissue dysplasia (hypermobility of the interatrial septum, additional chordae in the left ventricle cavity); the local and the global systolic function of the left ventricle is not impaired; type I diastolic dysfunction of the left ventricle; mitral insufficiency grade I; tricuspid insufficiency grade II;

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- ultrasound examination of the abdominal cavity organs: no ultrasonographic signs of pathological changes were detected in the liver, pancreatic gland and the bile ducts.

Prognosis

Is was shown that, in adult patients with late onset Pompe disease, not receiving enzyme replacement therapy, the mortality is higher than in general population [9]. The factors, improving the prognosis and slowing the progression of the disease, include the earlier diagnostics of the disease and prescribing enzyme replacement therapy.

DISCUSSION

The patient's way to receiving the pathogenetic therapy was approximately 20 years. The long-term period of diagnostics is related to the insufficient awareness and knowledge among the neurology physicians, which were following up the patient with the diagnosis of osteochondrosis. The late onset of the disease associated with "softer" mutations in the *GAA* gene and slower progression [10] also resulted in the delaying the process of diagnostics.

The findings detected in our patient upon the physical examination, namely, the muscular weakness syndrome, had to be differentiated with a number of diseases, and first of all, the impairment level needed to be determined — myogenic, neurogenic, synaptic or central. Taking into consideration the fact that the pyramidal syndrome (increased reflexes, pathological reflexes) was not reported in a patient, the central level of impairment of the neuromuscular system was immediately rejected. Our patient also did not have the pathological muscular fatigability symptom characteristic for the synaptic level of impairment. The absence of sensitivity disorders has called into question the neurogenic level, however, the damage of the motor neuron, as well as motor neuropathies, occur without impairing the sensitivity. In differentiating the neurogenic and the myogenic levels, electroneuromyography is helpful (with a proviso that the clinically weak muscles are examined). Electroneuromyography in cases of myogenic lesions has a number of limitations related to the mosaic pattern of muscle involvement for various myopathies, as well as to the atrophy of separate muscle fibers even within a single muscle [11, 12], which is why the movement unit potentials of the muscle, in which there are no objectively signs of strength decrease, can show normal pattern, in some cases even neurogenic, which can mislead the diagnosing specialist. The distribution of muscle weakness in our case and in the majority of cases described in the literature sources for late onset Pompe disease, has a specific pattern, characteristic by the predominant damage of the axial muscles, the proximal muscles of the upper and lower limbs, with the lower limbs being affected to a greater extent with intact strength in the feet [11, 13, 14]. In this clinical case, muscle weakness prevailed exactly in thigh muscles: the weak muscles were examined (the adductor and the quadriceps), in which single spontaneous activity and myogenic remodeling of the movement unit potentials were registered. The spontaneous activity in the affected muscles indicate the on-going process of denervation as a result of dying muscle fibers, in favor of which was the detected increase of blood CPK, however, the not so high CPK level and the activity of denervation in the muscles were indicating the slow course of the pathological process, which was also shown in other cases of late onset Pompe disease [13, 15, 16].

MRI of the muscles have confirmed the morphological remodeling of the thigh muscles. The myopathic syndrome, detected clinically and confirmed instrumentally, often does not receive nosological identification and remains a syndromal diagnosis. The tracked history of muscle weakness from the school years with slow progression indicates the possible genetic genesis of myopathy; the low rising CPK level and low activity of denervation count against the inflammatory damage of the muscles (polymyositis, dermatomyositis), the characteristic features of which include very high CPK levels and the activity of denervation; while the absence of the patient's relatives in the previous generation speaks for the autosomal-recessive mode of inheritance.

The accumulation diseases are divided into two main groups: with the predominant damaging of the nervous system (leukodystrophies) and with the predominant damaging of the muscular system (glycogenoses). In our case, the leading was the damage of the muscles. The myopathic syndrome in cases of glycogenoses, the number of which counts more than ten of types, is nosologically non-specific. The mode of inheritance in the majority of glycogenoses is autosomal-recessive. In case of type I (Gierke disease), the myopathic syndrome, unlike the late onset Pompe disease, is accompanied by hyperuricemia and hypoglycemia symptoms, by chronic renal failure, having an onset of the disease at the babyhood and being characterized by delayed growth and puberty [17]; in type III (Cori disease), besides hyperuricemia and hypoglycemia, hyperlipidemia

and hypercholesterolemia are observed [17]; in case of type IV (Andersen disease), patients show delayed development and the lethal outcome occurs due the hepatic failure to the 5th year after birth [18]; type V (McArdle disease) myopathy is accompanied by muscle pain, renal failure develops with a background of myoglobinuria along with hyperuricemia [19]. Thus, the late onset Pompe disease in adults differs from other glycogenoses by the absence of delayed physical development, hypoglycemia, hyperuricemia or myoglobinuria, however, its more credible differentiation is possible based on laboratory testing of the activity of the enzymes participating in the glycogen metabolism.

CONCLUSION

The confirmation of the diagnosis of late onset Pompe disease is conducted by means of using the laboratory and genetic tests studies. Dried blood spots can be sent to the genetic laboratory by any physician from any region of the country. To provide a possibility of receiving a chance for pathogenetic therapy for a patient, the physician should not only suspect the presence of a myopathic syndrome in a patient, but he should also be aware on the possibilities of sending dried blood spot samples for testing.

Insufficient positive dynamic changes in the patient status is related to the later onset of therapy, however, the absence of subjective aggravation of the symptoms indicates the stabilization of the status and the necessity of continuing the life-time pathogenetic enzyme replacement therapy for increasing the duration of the active phase period.

ADDITIONAL INFORMATION

Supplement 1. Trendelenburg gait, the patient experiences significant difficulties when standing up from the sitting position, from the lying position, from squat on haunches, using the Gower's maneuvers (weakness of axial muscles). doi: https://doi.org/10.17816/clinpract678919-4340943

Author contributions. *A.V. Yamshchikova* — diagnostic, search and analytical work, discussion of research results, writing the article text; *T.A. Bychkovskaya* — diagnostic work, discussion of research results, editing the article text. The authors made a substantial contribution to the conception of the work, acquisition, analysis, interpretation of data for the work, drafting and revising the work, final approval of the version to be published and agree to be accountable for all aspects of the work.

Consent for publication. The authors received written informed voluntary consent from the patient to publish personal data, including photographs (with face covering), in a scientific journal, including its electronic version (signed on 2025 April 12). The amount of published data is agreed with the patient.

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