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Difficulties in the Diagnostics of Transthyretin Amyloidosis with Polyneuropathy: a Clinical Case Description

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ABSTRACT

BACKGROUND: *In clinical practice, diagnosing of systemic transthyretin amyloidosis (ATTR-amyloidosis) with the impairment of the nervous and cardiovascular systems became possible due to the accessibility of genetic diagnostics and due to the growth of knowledge on this disease.* ***CLINICAL CASE DESCRIPTION:*** *A clinical case is presented of transthyretin amyloidosis, manifesting as polyneuropathy with significant neuropathic pain syndrome, with the development of severe vegetative insufficiency and with the impairment of the cardiovascular system in a 63 years old male. This clinical pattern, developing for 3 years, was associated with multiple encounters of the patient to various specialists — rheumatology physicians, neurosurgery specialists, endocrinologists, neurologists, internists, including surgical interventions in the spinal cord, carotid artery and cardiac vessels. The disease has lead to the development of severe cachexia and incapacitation of the patient.* ***CONCLUSION:*** *Taking into consideration that ATTR-amyloidosis manifests with the clinical signs of lesions in various organs and systems, and this is interpreted by the specialists within the framework of their specific field apart from the general etiology of the disease. The patient gets prescribed with multiple examinations and symptomatic medications, which causes a delay in the diagnostics and in setting the correct diagnosis. This clinical case describes the classical form of transthyretin amyloidosis, which, upon timely diagnostics, has its pathogenetic therapy.*

Keywords: *transthyretin amyloidosis; polyneuropathy; vegetative dysfunction; orthostatic hypotension; clinical case.*

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BACKGROUND

Transthyretin amyloidosis (ATTR-amyloidosis) is a rare disease with irreversibly progressive course due to the accumulation of the pathological protein — the transthyretin (TTR) in the peripheral nervous system, kidneys, heart, intestines and other organs [1–3].

The protein name “transthyretin” (transports thyroxine and retinol) is related to the transportation of the thyroid gland hormone thyroxin (T4) and retinol upon binding to the retinol-binding protein in the target organs for normal functioning. Up to 95% of transthyretin is synthesized by hepatic cells and secreted into the circulation [4, 5], with the remaining part being produced in the vascular plexus of the brain and in the pigmented epithelium of the eye retina and it circulates in the cerebrospinal fluid, not reaching the circulation. The TTR secretion begins in the process of embryonic development and continues throughout the entire life. The blood concentration of transthyretin is minimal in neonates, but with ageing, an increase develops in the levels of this protein with its gradual decrease after the age of 50 years [6].

The most common cause of ATTR-amyloidosis is the mutation in the *TTR* gene, resulting in an impaired secondary and tertiary structure of the TTR protein: in cases of the pathological type, transthyretin does not reproduce its usual tetrameric form, but forms the amyloid fibrils. The accumulated TTR-protein affects the operation of the cell with its further death [1, 3].

The first clinical course of familial transthyretin amyloid polyneuropathy was described in 1952 by the Portuguese Neurologist C. Andrade in patients from Northern Portugal, and later on, due to the development

Сложности в диагностике транстиретинового амилоидоза с полинейропатией: клинический случай

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АННОТАЦИЯ

Обоснование. *В клинической практике диагностирование системного транстиретинового амилоидоза (ATTR-амилоидоз) с поражением нервной и сердечно-сосудистой систем стало возможным благодаря доступности генетической диагностики и увеличению знаний о данной патологии.* ***Описание клинического случая.*** *Представлен клинический случай транстиретинового амилоидоза, проявляющегося полинейропатией с выраженным нейропатическим болевым синдромом, развитием грубой вегетативной недостаточности и поражением сердечно-сосудистой системы у мужчины 63 лет. Данной клинической картине, развивавшейся в течение 3 лет, сопутствовали многочисленные обращения пациента к различным специалистам — ревматологам, нейрохирургам, эндокринологам, неврологам, терапевтам, в том числе оперативные вмешательства на спинном мозге, сонной артерии и сосудах сердца. Заболевание привело к выраженной кахексии и инвалидизации пациента.* ***Заключение.*** *ATTR-амилоидоз проявляется поражением различных органов и систем, и это воспринимается врачами-специалистами в рамках своей области в отрыве от общей картины заболевания. Пациенту назначаются многочисленные исследования и симптоматические препараты, что вызывает задержку в диагностике и постановке правильного диагноза. Данный клинический случай описывает классическую форму транстиретинового амилоидоза, которая при своевременной диагностике имеет патогенетическое лечение.*

Ключевые слова: *транстиретиновый амилоидоз; полинейропатия; вегетативная дисфункция; ортостатическая гипотензия; клинический случай.*

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of genetic diagnostics, this neurodegenerative disease was diagnosed worldwide [1, 7].

The incidence of developing the transthyretin amyloidosis is variable depending on the regions, even among the members of one family. Taking into consideration the incomplete penetrance, the individuals bearing the pathological gene can live long enough without clinical manifestations of the disease [5]. The average life expectancy from the moment of clinical manifestations until the lethal outcome for TTR-amyloidosis is 15 years, but these ranges can vary depending on the genotype, the age of disease onset, the clinical signs and the place of residence [2].

TTR-amyloidosis represents in two forms [3] — the acquired one (caused by the abnormal accumulation of wild type transthyretin) and the hereditary form (more than 140 variants of mutations in the *TTR* gene).

The clinical manifestations of transthyretin amyloidosis include the polyneuropathy with severe pain syndrome; the carpal canal syndrome; the dysfunction of the vegetative nervous system of unclear etiology (erectile dysfunction, orthostatic hypotension, impaired motor activity of the gastrointestinal tract, cardiac rhythm disorder); cardiac impairment with an enlargement of the walls in the right and left ventricles in the absence of arterial hypertension and combined with orthostatic hypotension [1, 3, 7].

Predominantly, the first manifestation of TTR-amyloidosis is the polyneuropathic syndrome, less frequently — the vegetative disorder. In 18% of the

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journal of clinical practice 2025 Vol 16 №2

cases amyloidosis affects the heart, which results in rhythm abnormalities, requiring the installation of the pacemaker [1, 3].

TTR-amyloidosis was considered an incurable disease until the development of the first liver transplantations in 1990-s [1]. As of today, the pathogenetic therapy of transthyretin amyloidosis for the purpose of stabilizing the TTR-tetramers is carried out using the medicinal agents Diflunisal and Tafamidis [5]. Diflunisal, being a drug product from the group of non-steroid anti-inflammatory medications, affecting the foci of T4 binding at the TTR, slows down the process of amyloidogenesis [5, 8], however, taking into consideration the negative effects on the side of the gastrointestinal tract, for 57% of the patients, this medicine is currently not approved for use (research works are pending) [8]. Tafamidis stabilizes the TTR tetramer by means of binding to the thyroxin-binding site of the TTR, both the wild type and the mutated one [8, 9]. In patients with the manifestation of clinical signs at the age of <50 years, Tafamidis was decreasing the risk of mortality comparing to the risk for the patients not receiving therapy by 91%, while among the individuals with the onset of the disease at ≥50 years of age — by 82% [9].

CLINICAL CASE DESCRIPTION

Patient info

Patient named M., aged 63 years, a retired resident of the city of Tyumen, was admitted in June 2024 to the Neurology Department of the State Budgetary Healthcare Institution of the Tyumen Oblast "Regional Clinical Hospital No. 1" (RCH No. 1 of the city of Tyumen) with the complaints of weakness and pain in the muscles of the limbs, pain in the vertebral column, dizziness, generalized weakness, frequent loose stools and impaired erectile functions.

Case history. From 2020 (at the age of 59 years), the patient started noting weakness and prickling in his upper and lower limbs, intensive pain in the limbs and in the vertebral column, decreased blood pressure (down to 90/60 mm.Hg.) along with erectile dysfunction. From November 2021, headaches and dizziness have also developed.

Due to the persisting pain syndrome, on 25.01.2022 the patient was examined by the Rheumatologist, no abnormalities were detected. One and a half year from the moment of the clinical manifestations, the patient had lost 20 kg of body weight. From February 2022, shortness of breath started developing upon physical activity: the coronary angiography on 03.08.2022 has shown no abnormalities. In July 2022, carotid endarterectomy was arranged on the right side due to the hemodynamically significant stenosis of the internal carotid artery — down to 85%.

From 19.09.2022 until 29.09.2022, the patient was staying at the Neurosurgery Department of the Federal Center for Neurosurgery of the city of Tyumen with the complaints of pain and numbness in the upper and lower limbs, pain in all the segments of the vertebral column. Objective assessment has revealed the "gloves and socks" hypesthesia, significant tremors in the distal segments of the upper limbs. Clinical hematology panel: mild anemia (Red blood cells 3.63×10^{12}/l, hemoglobin 108 g/l). Magnetic resonance tomography of the cervical, thoracic and lumbar spine on 19.09.2022: arthrosis of the facet joints, mild central stenosis at the level of C6–C7, moderate — at the levels C4–C6 and Th10–Th11. Moderate bilateral foraminal stenosis at the level of C4–C7. Electrocardiography was conducted on 19.09.2022: sinus rhythm with a heart rate of 77 bpm, left anterior fascicular block, signs of loading in the right atrium. On 23.09.2022, a decision was drawn up on arranging the temporary epidural electrostimulation of the spinal cord at the level of C3–C5. The postoperative period was unremarkable (no complications), weakly positive effect was reported.

From 02.11.2022 until 11.11.2022, the patient was receiving scheduled treatment at the Cardiology Department of the Tyumen RCH No. 1.

Laboratory and instrumental diagnosis

On 07.11.2022, the transluminal balloon angioplasty and stenting of the coronary artery were performed. The postoperative period was unremarkable (no complications). The clinical hematology panel was showing persisting anemia (Red blood cells 3.41×10^{12}/l, hemoglobin 92 g/l). Echocardiography on 11.12.2022: atherosclerosis of the aorta, insignificant myocardial hypertrophy of the left ventricle, insignificant amount of fluid within the pericardial cavity.

From 27.12.2022 until 04.01.2023, the patient underwent in-patient treatment at the Endocrinology Department. Upon visual examination, the body weight was 40 kg, height — 168 cm, body mass index (BMI) — 14.17 kg/m^2. The blood cortisol concentration was less than 1 μg/dl (ref. range: 3.7–19), adrenocorticotropic hormone — 2.3 pg/ml (ref. range: 7.2–63.8). The diagnosis set was the secondary insufficiency of the adrenal cortex, the intake of Prednisolone was recommended at a dosage of 5 mg daily.

On 08.01.2023 — transported by the ambulance crew to the Tyumen RCH No. 1 with arterial hypotension (blood pressure — 50/35 mm.Hg.) and complaints of severe weakness, prickling and numbness in the upper and lower limbs, severe dizziness and shortness of breath on minimal physical loading.

Clinical hematology panel: Red blood cells 3.65×10^{12}/l, hemoglobin 108 g/l with a decrease of serum iron concentration down to 7.7 mmol/l. The blood biochemistry panel and the clinical urinalysis were unremarkable. The C-reactive protein level was normal. The parathyroid and the thyroid-stimulating hormones, the Free T4, the luteinizing and the follicle-stimulating hormones were normal, as well as the levels of cortisol. Decreased blood testosterone was found — down to 3.1 ng/ml. Upon the ultrasound examination of the thyroid gland, kidneys and adrenal glands — no abnormalities were detected. Electrocardiography on 10.01.2023: sinus rhythm with a heart rate of 89 bpm, left anterior fascicular block, signs of loading in the right atrium, signs of hypertrophy in the left ventricle. Electroneuromyography on 12.01.2023: signs of sensory-motor polyneuropathy in the lower limbs, predominantly of axonal type. X-ray densitometry on 12.01.2023: the lumbar segment of the vertebral column and the femoral neck show decreased mineral density down to the level of osteoporosis (T-Student's test -2.9 SD and -3.3 SD respectively). Ultrasound duplex scanning of the brachiocephalic arteries on 13.01.2023: atherosclerosis of the brachiocephalic arteries, stenosis down to 45–50%. Computed tomography of the chest cavity organs: signs of insignificant mixed-type emphysema. The patient was discharged in generally satisfactory status with stable hemodynamic parameters.

On 20.06.2023, due to the persisting pain syndrome in the back and limbs along with the numbness in the upper and lower limbs, the patient was repeatedly admitted to the Neurosurgery Department of the Federal Center for Neurosurgery of the city of Tyumen, where he once again underwent an implantation of the system intended for epidural electrostimulation of the spinal cord at the level of C3–C5 vertebral bodies.

Conclusion on the electroneuromyography examination of the upper and lower limbs (self-encounter due to the persisting numbness in the limbs): signs of significant motor-sensory axonal-demyelinating polyneuropathy of the limbs with an accent in the lower ones. The amplitude of the motor and sensory responses is significantly decreased (>96%). The rate of conduction of excitation in the peripheral nerves is decreased in the upper (by 25–30%) and in the lower (35–50%) limbs.

Physical diagnostics. Body weight 40 kg. Height 168 cm. BMI 14.17 kg/m^2. General status — satisfactory. Heart rate — 62/minute. The pulse is rhythmic. Blood pressure in the right arm — 86/60 mm.Hg. in the horizontal position, 55/40 mm.Hg. in the vertical position, in the left arm — 74/50 mm.Hg. in the vertical position. The heart tones are muffled but rhythmic.

Neurological status. The critical thinking is intact. The oculomotor nerves show no abnormalities. The face is symmetrical. Primitive oral reflexes: snout reflex. The soft palatine and the gag reflexes are intact. The muscle strength in the upper limbs — proximally 4 points, distally — 2–3 points. The muscle strength in the lower limbs is proximally 3–4 points, distally — 2 points. The muscle tone in the limbs is decreased. Atrophy in the muscles of the hypothenar, thenar, trapezius muscles of the shoulder joint, in the thigh and shin muscles. Fascicular tremors were revealed in the muscles of the shoulder girdle. The reflexes in the upper limbs are active in the biceps and triceps muscles, no carpal reflexes were observed. The reflexes in the lower limbs: knee and Achilles tendon — none on both sides. Pathological plantar reflexes — absent. Romberg stance test shows sensitive ataxia. Coordination tests — an intention tremor was detected on both sides. Hypoaesthesia up to the upper third of the forearm, down to the lower third of the thighs. Paraesthesias in the lower limbs. The functions of the pelvic organs are properly controlled.

Provisional diagnosis

Axonal type polyneuropathy, unspecified.

Genetic examination

Taking into consideration the clinical signs and the case history, the patient had his blood sample drawn for conducting the genetical testing for the purpose of ruling out the transthyretin amyloidosis, and on 30.04.2024, using the Sanger sequencing method, a pathogenic mutation was detected in exon 2 of the *TTR* gene (Nm_000371.4) c.148G>A (p.Val50Met).

For the purpose of ruling out the amyloidosis with light chain amyloid (AL-type of amyloidosis), 24-hour samples of blood and urine were tested (21.06.2024): the concentration of immunoglobulins are within the normal ranges, no monoclonal secretion detected. Scintigraphy of the myocardium was arranged (21.05.2024): accumulation of the amyloid was shown in the heart, malignancy degree (Grade) III (Fig. 1).

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journal of clinical practice 2025 Vol 16 №2

Main diagnosis

Transthyretin amyloidosis with polyneuropathy and amyloid lesions in the heart, grade III, severe sensitive ataxia, severe neuropathic pain syndrome, moderate flaccid tetraparesis.

Concomitant diagnoses. Secondary insufficiency of adrenal cortex. Orthostatic hypotension. Significant body weight deficit (BMI 14.17 kg/m^2). Adenoma of the prostate gland. Degenerative-dystrophic changes in the vertebral column, thoracalgia syndrome, lumboischialgia with stable bilateral pain syndrome, incomplete remission and chronic recurrent course. S/p implantation of the system for epidural electrostimulation of the spinal cord on 20.06.2023. Carotid endarterectomy of the internal carotid artery on 29.07.2022. Ischemic heart disease. Atherosclerosis of coronary arteries. Transluminal balloon coronary angioplasty with stenting the right coronary artery on 07.11.2022. Hypertensive disease stage III, risk 4, controlled. Chronic cardiac failure grade I, functional class II.

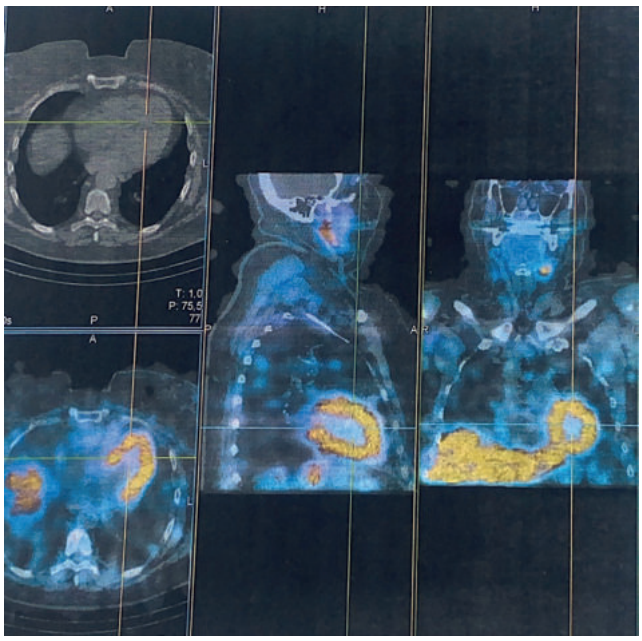

Fig. 1. Scintigraphy of myocardium: accumulation of amyloid in the heart, grade III of malignancy (Grade).

Follow-up and outcomes

The genetic testing of blood samples from the three sons of the patient shows that none of them is carrying a pathological variant c.148G>A (p.Val50Met) in the *TTR* gene, detected in their father.

The patient has received recommendations on taking the medicinal product Tafamidis 61 mg once daily, not crushing the pill and not cutting it, not related to the intake of meals.

DISCUSSION

Transthyretin amyloidosis is a multisystemic disease associated with the deposition of amyloid in the neural tissue, in the heart, the kidneys, the eyes and the gastrointestinal tract. Taking into consideration the polysystemic origin of the disease, amyloidosis often mimics the disease of the internal organs, due to which the clinical signs with the predominance of one or another complaints are investigated by various specialists (neurologists, cardiologists, endocrinologists etc.), which results in delayed setting the correct diagnosis, the progression of the disease with the development of decompensation in the organs and systems, delaying the initiation of the pathogenetic therapy and, as a result, the incapacitation of the patient with decreasing the quality of his/her life. In the endemic regions (Portugal, Sweden, Japan, Brazil, Cyprus, Majorca, Bulgaria), transthyretin amyloidosis is found with an occurrence rate of 1:1000, while in cases of hereditary ATTR-polyneuropathy, known in 36 countries of the world — in 1:1 000 000 of the population [10]. Setting the correct diagnosis in patients with amyloidosis takes an average of approximately three years [2], which is related to the absence of hereditary history, to multiple and non-specific clinical manifestations. The most frequent neurological diagnoses, which are set for the patients with ATTR-amyloidosis, include polyneuropathy of unspecified origin, chronic inflammatory demyelinating polyneuropathy, lateral amyotrophic sclerosis, degenerative-dystrophic changes of the vertebral column with radiculopathy and Parkinson disease [2].

Early diagnostics of ATTR-amyloidosis allows for prescribing the pathogenetic therapy (according to the vital indications) before the development of irreversible incapacitating changes in the organs and systems, the therapy that is aimed at preventing the accumulation of amyloid. In Russia currently there is a registered pathogenetic therapy medicine with proven efficiency — Vyndaqel (Tafamidis)[1], which selectively binds to the transthyretin protein, causing its stabilization and preventing amyloid deposition [11], by this significantly slowing the progression of neurological symptoms, preventing the loss of body weight in patients with ATTR-polyneuropathy, which is confirmed by the results of numerous observations

[1] Instructions on the medical application of Vyndaqel drug product. Access mode: https://grls.rosminzdrav.ru/GRLS.aspx?RegNumber=&MnnR=&lf=&TradeNmR=Виндакель&OwnerName=&MnfOrg=&MnfOrgCountry=&isfs=0&regtype=1%2c6&pageSize=10&token=7e96bd09-3b25-4dcf-a2b1-79aa4abc9c58&order=Registered&orderType=desc&pageNum=1

from foreign authors [8, 9, 11, 12]. In the presented clinical case, what calls attention to itself is practically four years of delayed period from the development of polyneuropathy symptoms and erectile dysfunction to setting the clinical diagnosis. The patient was supervised by the endocrinologists, rheumatologists, cardiologists, neurologists, he had twice underwent an invasive implantation of epidural electrodes due to his pain syndrome. Thus, the patient underwent multiple examinations and was repeatedly operated.

The provided observation demonstrates the irreversibly progressing type of the course of transthyretin amyloidosis with the dysfunction of the vegetative nervous system, expressed as the development of orthostatic hypotension, erectile dysfunction, impaired motor functions of the gastrointestinal tract with a background of completely intact cognitive functions. The accumulation of the modified amyloid has lead to the deficit of the body weight (BMI 14.34 kg/m^2), to the severe pain syndrome in the limbs, to difficulties in unassisted movement and to severe vegetative disorders. At the same time, the timely conducted genetic testing, setting the correct diagnosis and prescribing proper therapy could prevent the incapacitation of the patient.

CONCLUSION

Practicing physicians need to remember about ATTR-amyloidosis, the timely genetic diagnostics of which allows for prescribing the pathogenetic therapy, slowing down the progression of the disease and significantly improving the quality of life in a patient.

ADDITIONAL INFORMATION

Author contributions. *E.S. Ostapchuk:* review of publications on the topic of the article, scientific editing of the article; *O.P. Glinin:* description of the clinical case, review of publications on the topic of the article; *Yu.V. Alekseeva:* treatment of the patient, correction of the handwritten part of the 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 the face covered), in a scientific journal, including its electronic version (signed on 2024 July 8). The volume of published data was agreed upon with the patient.

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