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Some clinical manifestations of Fabry disease, aspects of diagnosis and treatment

Dzhanibekova A.R.[1], Uzdenov M.B.[1], Dzhanibekova L.R.[1,2], Bogatyreva Z.R.[3], Mutchaeva F.M.[1], Tambieva Z.N.[1]

[1] North Caucasus State Academy, Medical Institute, Cherkessk, Russia.
[2] Karachay-Cherkess Republican Clinical Hospital, Cherkessk, Russia.
[3] Stavropol Regional Clinical Hospital, Stavropol, Russia.

AUTHORS

Asiyat R. Dzhanibekova*, PhD, MD, Associate Professor, Department of Internal Medicine, Dean of the Faculty of International Students, Medical Institute, North Caucasus State Academy; Cardiologist, Functional Diagnostics Physician, Cherkessk, Russia. ORCID: 0000-0002-4719-9980

Marat B. Uzdenov, PhD, MD, Director of the Medical Institute, Associate Professor, Head of the Department of Topographic and Pathological Anatomy with a Course in Operative Surgery, North Caucasus State Academy, Cherkessk, Russia. ORCID: 0000-0002-0077-9013

Leila R. Dzhanibekova, PhD, MD, Associate Professor, Department of Internal Medicine, Medical Institute, North Caucasus State Academy; Cardiologist, Head of the Emergency Cardiology Department, Karachay-Cherkess Republican Clinical Hospital; Chief Freelance Cardiologist of the Ministry of Health of the Karachay-Cherkess Republic, Cherkessk, Russia. ORCID: 0009-0003-6535-2744

Zamira R. Bogatyreva, Cardiologist, Stavropol Regional Clinical Hospital, Stavropol, Russia. ORCID: 0000-0002-0076-3276

Fatima M. Mutchaeva, Assistant, Department of Internal Medicine, Medical Institute, North Caucasus State Academy, Cherkessk, Russia. ORCID: 0009-0006-0022-8250

Zaira N. Tambieva, Assistant, Department of Internal Medicine, Medical Institute, North Caucasus State Academy, Cherkessk, Russia. ORCID: 0009-0007-2408-3997

Mitochondrial diseases are considered among the most common genetic metabolic disorders. A special group consists of lysosomal storage diseases, one form of which is Fabry disease (FD). This is a progressive orphan hereditary metabolic disorder linked to the X chromosome, with a mutation occurring in the GLA gene encoding the en-

* Corresponding author. Tel./Тел. +7 9280110271. E-mail: asia.janibekova@mail.ru

zyme α-galactosidase A, resulting in decreased or absent activity of this enzyme, leading to abnormal accumulation of glycophospholipids and metabolic disturbance.
Objective of this review article is to examine current literature on the pathogenesis, factors playing an important role in the disease pathogenesis, diagnostic algorithms and treatment of FD.
Materials and methods. This review article used works by Russian and foreign authors published on internet platforms and in print over the past 10 years.
Results. The review article discusses current literature on clinical manifestations, factors important in disease diagnosis, modern aspects of diagnosis and principles of FD treatment. Recent studies have also added information on the epidemiology of FD.
Conclusion. Diagnosis and treatment of FD have evolved significantly in recent years due to improved patient selection algorithms, progress in laboratory and instrumental diagnostics of the pathology, and increased accessibility of these methods for timely identification of clinical cases. However, a comprehensive understanding of the etiology, pathogenesis and consequently the clinical manifestations of the disease has still not been achieved, leading to late diagnosis and lack of timely pathogenetic therapy. Therefore, further research is needed to elucidate the factors involved in early diagnosis of the disease in patients with FD.
Keywords: hereditary metabolic disorder, lysosomal storage diseases, Fabry disease, epidemiology, α-galactosidase A, enzyme replacement therapy.

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Introduction

In recent years, there has been growing interest in new nosological forms of hereditary monogenic pathology belonging to the group of lysosomal storage disorders (LSDs), with increasing attention being paid to Fabry disease (FD), also known as Anderson-Fabry disease.

FD is a rare, progressive, inherited metabolic disorder caused by mutations in the GLA gene located on the X chromosome, which encodes the enzyme α-galactosidase A (α-Gal A). Consequently, there is an absence or deficiency of its activity, leading to the abnormal accumulation of two types of phospholipids: primarily globotriaosylceramide and its derivative globotriaosylsphingosine in the lysosomes of cells in various tissues (vascular endothelium, kidneys, heart, central nervous system, peripheral nervous system, etc.), as well as in bodily fluids [1].

These changes result in multisystem manifestations of the disease, leading to progressive organ damage and, consequently, a reduced quality and length of life. In the absence of appropriate treatment, death can occur at the age of 40-50 years due to cardiovascular, cerebrovascular complications, and renal failure.

At the same time, this diagnosis is not always established in a timely manner, leading to late diagnosis and the absence of pathogenetic therapy. Considering the large volume of available information and the numerous contentious issues surrounding FD, there is a need to analyze the literature data.

Objective—To summarize current data regarding the mechanisms of disease onset, clinical manifestations, diagnostic criteria, features of the course, and treatment of FD based on studies conducted in recent years.

Materials and methods

This review article utilized the work of Russian and foreign authors and involved a systematic search in the PubMed and e-library databases, on internet platforms, and in print sources using the following keywords: "Fabry disease," "alpha-galactosidase A," "lysosomal storage disease," "Globotriaosylceramide," "Anderson-Fabry disease," "diffuse angiokeratoma of the trunk" for the period from January 1, 2015, to January 1, 2026. Based on the search results, consensus documents, meta-analyses, scientific articles, and clinical cases were analyzed. Materials, studies, or articles that did not meet quality standards were excluded.

Results

Epidemiology. In terms of prevalence, FD ranks second among LSDs after Gaucher disease. Prevalence rates vary widely and are directly dependent on the disease phenotype. Foreign authors report the following rates for patients with the classic phenotype: from 1 in 117,000 to 1 in 476,000 in the general population [2]. Moreover, a predominance in prevalence is observed in males: 1 in 40,000 and 1 in 60,000 [3]. Considering the late-onset phenotypes, a higher actual prevalence of this pathology is clearly suggested. For example, according to a European multicenter study conducted by Elliott et al., pathogenic galactosidase mutations were identified in 0.5% of 1,386 patients with hypertrophic cardiomyopathy (HCM). According to Petrukhina A.A. et al., as of 2024, 230 clinical cases of FD were registered in Russia [4]. In another study by Eng C.M., Germain D.P., et al., the frequency of pathogenic mutations during screening was 1% [5]. Furthermore, interesting data emerged from screening studies in Italy, where the frequency of GLA gene mutations reached 1 case per 8,800 newborns [6]. In Taiwan, a similar study identified 1 case per 1,600 newborn males. It has been established that the average age of diagnosis for men and women is 24 and 31 years, respectively. At the same time, undiagnosed or untreated typical FD can reduce life expectancy by 15 years in men and 20 years in women [7].

Literature data were analyzed regarding the frequency of FD in various conditions. Thus, FD was found in 4.2% of men and 2.15% of women with early stroke, in 0.9% and 3.9% of men and women with HCM of unknown origin, and in 0.33% and 0.10% of men and women with end-stage renal disease, respectively [8].

Classification of the disease. Two forms of the disease are distinguished: the classic, or typical form, and the non-classic form, also known as the late-onset form [4, 8].

In the first case, patients with the classic phenotype may exhibit signs and symptoms of FD from early childhood, which are polyorganic in nature. These signs include neuropathic pain, verticillata cornea, gastrointestinal symptoms, hypohidrosis, and angiokeratoma. Delayed manifestations of the disease include chronic kidney disease (CKD) with renal failure, HCM, cardiac rhythm and conduction disturbances, hearing loss (bradyacusia or hypoacusia), and cerebrovascular accidents. This variant is characterized by the absence or significant reduction (less than 3% of the expected value) of α-Gal A activity.

In the second variant, signs and symptoms of FD develop at a later age, representing a milder course of the disease. Simultaneous involvement of all the aforementioned target organs may be insignificant or absent altogether, but there is often predominant and typically severe involvement of a single organ. In the non-classic variant, FD more frequently affects the brain, heart, or kidneys. This variant is characterized by a less significant reduction in α-Gal A levels. However, cardiac involvement is one of the most common clinical manifestations in patients with both variants of FD.

Clinical Manifestations of Fabry Disease

FD is "multifaceted" and presents with diverse manifestations even among members of the same family, meaning the clinical picture ranges from a complete absence of symptoms or a mild form to a severe one (Fig. 1). The spectrum of possible complaints is quite varied and depends on several factors, such as the disease form, individual characteristics of the disease course, gender, etc.

The disease is progressive in nature, and, as a rule, without specific treatment, death occurs in the 4th-5th decade of life due to renal failure or cardiovascular complications [9].

Cardiovascular Manifestations. It has been established that myocardial damage is a prognostically unfavorable and frequent manifestation of FD [11]. According to statistics, cardiac involvement is the leading cause of death in 34% of men and 57% of women [8, 11]. The mechanisms of cardiovascular system damage result from the deposition of glycosphingolipids in cardiomyocytes, endothelial and smooth muscle cells of vessels, endocardial cells, conducting tissue, and valvular fibroblasts. This leads to the formation of left ventricular hypertrophy (LVH), diastolic dysfunction, myocardial ischemia, and the occurrence of cardiac rhythm and conduction disturbances [8, 12].

According to research data, screening for FD among over 5,400 patients with LVH and/or HCM of unclear etiology revealed a frequency of pathogenic mutations of 0.93% and 0.90%, respectively [13]. Other manifestations of this disease are rhythm and conduction disturbances. Manifestation of coronary

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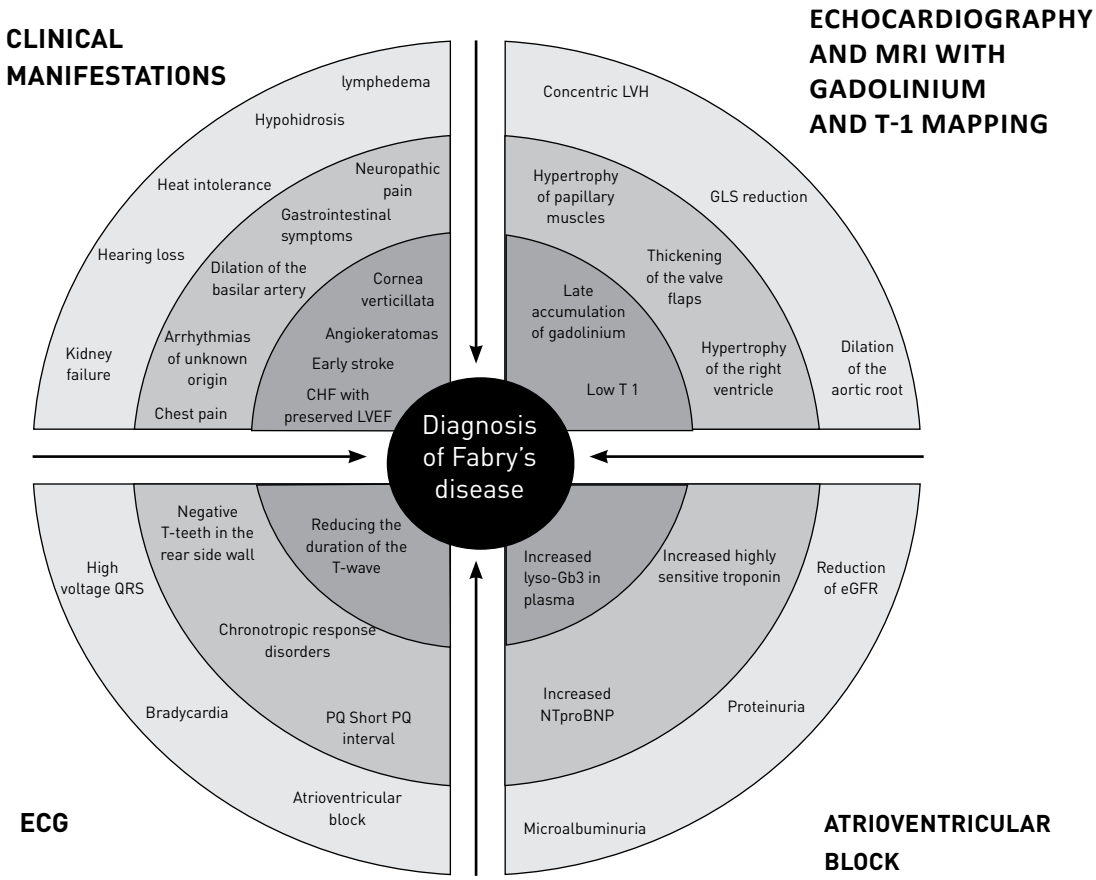

Fig. 1. Clinical manifestations and results of laboratory and instrumental studies that may indicate FD [10]. Darker shading indicates more characteristic signs of the disease

artery disease in the form of myocardial infarction occurs in 2% of patients with FD, while angina pectoris occurs in 13–20% of patients, often in combination with LVH [8, 13]. Valve involvement is also frequently observed in FD but has no particular clinical significance. As mentioned above, in the non-classical form with monosymptomatic course, cardiac involvement may be the sole manifestation of FD.

Renal involvement. Kidney damage can occur either as an isolated manifestation in the non-classic variant of FD or in conjunction with other syndromes in the classic variant. Renal involvement most often develops during adolescence and manifests as a nephritic urinary syndrome, potentially progressing to nephrotic syndrome and declining renal function up to end-stage chronic renal failure (CRF), which is one of the leading causes of death in patients [14].

Gastrointestinal manifestations. Clinical manifestations of GI tract involvement include non-specific symptoms such as decreased appetite, weight loss, intermittent cramping abdominal pain, bloating, unstable stools, nausea, and vomiting. The onset of gastroenterological manifestations of FD is more often observed in childhood and adolescence [8, 15].

Nervous system involvement. Damage to both the peripheral and central nervous systems is possible. Polyneuropathies, resulting from peripheral nervous system damage, are relatively common clinical syndromes in FD. They occur in 60–80% of patients, manifesting as chronically severe, sometimes debilitating neuropathic pain in the extremities, presenting as acroparesthesias, allodynia, and hyperpathia. Characteristic types of chronic pain include burning, shooting, and stabbing. The pathogenesis of neuropathy involves two mechanisms of neuropathic pain: central and peripheral. In the first case, damage to the thalamus occurs due to calcification in FD. In the formation of peripheral neuropathic pain, accumulation of globotriaosylceramide occurs in nerve axons, dorsal root ganglia, and vasa nervorum. Central nervous system involvement manifests as vascular events such as transient ischemic attacks, ischemic or hemor-rhagic stroke occurring between the ages of 20 and 50. Stroke can be a form of monosymptom-

atic manifestation of the atypical form of FD. The prevalence of stroke is about 6.9% and 4.3% in men and women, respectively. Chronic cerebral ischemia can lead to vascular dementia, including memory decline, impaired concentration, and behavioral disturbances [8, 16].

Sweating disorders. Accumulation of globotriaosylceramide in sweat glands, vessels, and autonomic nerve fibers leads to hypohidrosis or anhidrosis in the majority of cases.

Lymphatic system involvement. In some cases, lymphedema (pathological accumulation of protein-rich fluid in the interstitial space due to impaired lymph transport, accompanied by an increase in the volume of the affected organ) may be observed. Lymphedema can result from congenital or acquired impairment of lymph transport through lymphatic vessels due to glycolipid deposition in the lymphatic vessel walls [8, 17]. Scientific literature describes cases of lymphedema complicated by trophic disorders, leading to ulceration of the skin.

Otorhinolaryngological manifestations. Involvement of the auditory and vestibular systems may manifest as tinnitus (unilateral or bilateral), sensorineural hearing loss, and dizziness [8, 18].

Skin manifestations. Angiokeratomas are a manifestation of skin involvement in patients with FD. These are well-demarcated papules, red, dark red, burgundy, purple, or blue-black in color, ranging from 0.2 to 1.0 cm in size, predominantly located symmetrically in the groin area, on the buttocks, thighs, around the navel, and on the palms. Initially, angiokeratomas appear in childhood, are lighter, flatter, and more compressible, but with age their number and size increase, and they become darker, harder, and more elevated above the skin surface. Cases of angiectasias in the oral mucosa and conjunctiva have been described. Skin manifestations are not obligatory clinical signs.

Ophthalmological manifestations. Lens and corneal involvement occurs in more than 50% of patients with FD. A typical symptom is corneal opacity in a whorl-like pattern (in 70–90% of patients). Occasionally, lens opacities such as posterior radial subcapsular cataract, the so-called "Fabry cataract," and bilateral anterior capsular and subcapsular cataracts may occur, but these are not pathognomonic signs [19].

External features. External signs resemble those of acromegaly—prominent supraorbital ridges and frontal bosses, a protruding lower jaw, thickened lips, and a depressed nasal bridge are often observed in hemizygous males.

Features of the clinical picture in childhood and adolescence. Most symptoms of FD manifest during adolescence and young adulthood; however, verifying the diagnosis of FD in children is difficult due to the non-specific nature of the disease syndromes. For example, GI manifestations in this disease can only serve as an indirect marker due to their low specificity. Angiokeratomas in childhood are often solitary and may occur in atypical locations such as the chest, oral mucosa, and ears.

Manifestations of cardiovascular system involvement are also non-specific, presenting as periodic non-significant decreases and/or increases in blood pressure, asymptomatic or oligosymptomatic arrhythmias mimicking autonomic dysfunction. Timely diagnosis of renal involvement in FD is challenging, as this condition may manifest only as transient microalbuminuria [8, 20]. Rarer, less specific manifestations of FD in this age group include glossitis, granulomatous cheilitis, osteopenia, osteoporosis, hypothyroidism, thickening of the terminal phalanges, anemia, and respiratory disorders.

Diagnosis of the disease. In establishing a diagnosis of FD, medical history data and clinical-laboratory (molecular-genetic and biochemical) research methods are important. Verification of the classic variant of FD relies on the combination of multi-organ involvement, while verification of atypical variants is possible through screening of risk groups. It is important to note that FD should be suspected in all patients with young-onset stroke and LVH of unknown etiology.

Laboratory diagnostic tests

1. **Enzyme assay:** Determination of alpha-galactosidase A (AGAL) activity in dried blood spots on filter paper or in blood leukocytes for all male and female patients; however, in about one-third of female patients with FD, this indicator may be within the normal range.

2. **Identification of mutations** in the gene encoding AGAL for men with reduced enzyme activity and for women with clinical signs of FD using Sanger sequencing of all exons and flanking intronic regions.

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This method is also necessary for identifying the familial GLA gene mutation in relatives of the proband for prenatal and preimplantation diagnosis, as well as for identifying heterozygous female carriers of FD.

3. **Quantitative determination** of globotriaosylsphingosine in dried blood spots or blood plasma is performed for all patients, both for biochemical confirmation of the FD diagnosis and for biochemical monitoring of therapy, given that this metabolite does not reach normal values in patients with FD.

4. Conducting laboratory tests according to clinical guidelines for managing patients with CRF (at least once every 6 months) and HCM when clinical signs of renal and cardiac involvement are present.

5. For all patients, regular (at least once every 6 months) determination of protein and albumin levels in urine and estimation of glomerular filtration rate are recommended to detect renal pathology.

6. Biopsy and pathological examination of kidney and heart tissues are recommended only in complex differential diagnostic cases [8, 21].

Instrumental diagnostic tests

To detect LVH, electrocardiography, echocardiography and Doppler echocardiography, and MRI are indicated. MRI of the brain is also recommended to detect possible involvement of the cerebral white matter (leukoaraiosis), gray matter (calcification of the posterior thalamus), ischemic foci, intracerebral hematomas, and vascular malformations. Additionally, tonal audiometry every 3 years is recommended to detect hearing loss.

The patient's examination plan also includes consultations with a cardiologist (at least once every 6 months) and an ophthalmologist (at least once every 3 years) [8].

Treatment principles

Treatment includes pathogenetic, symptomatic, and surgical approaches. Pathogenetic therapy includes enzyme replacement therapy and chaperone therapy. Other treatment methods are under clinical trial development, such as substrate reduction therapy, other enzyme replacement agents, and gene therapy.

Pathogenetic therapy. Pathogenetic therapy includes enzyme replacement therapy, which is currently represented by two drugs on the Russian pharmaceutical market: agalsidase alfa and agalsidase beta. The efficacy of therapy was evaluated in over 48 prospective studies, which showed a positive treatment effect, including reduced myocardial mass, decreased Cb3 levels in kidney tissues, reduced neuropathic pain, improved hearing, and enhanced quality of life. The use of a chaperone (migalastat), which is currently not registered in the Russian Federation, is also being considered. New treatment options for substrate-reducing therapy include lucerastat and venglustat, which are in Phase 2-3 trials. New pegylated forms of α-Gal A (pegunigalsidase alfa) and gene therapy agents are also being developed [9, 22]. The success of therapy directly depends on the disease form, timing of treatment initiation, drug dosage, potential development of antibodies to drug components, and several other factors.

Symptomatic therapy. Symptomatic therapy includes pain management and treatment of multisystem pathology according to current clinical guidelines. Symptomatic therapy may be prescribed by a medical commission decision [23]. For example, for pain management, the use of analgesics is recommended, along with eliminating or limiting provoking factors such as intense physical activity, temperature fluctuations (cold and/or hot weather), etc. For patients with cardiovascular complications, antiplatelet agents, anticoagulant, antiarrhythmic, and antihypertensive therapy are recommended. For patients developing gastrointestinal complications, the use of proton pump inhibitors or H_2-histamine receptor blockers is permitted. In the presence of proteinuria and arterial hypertension syndrome, the addition of angiotensin-converting enzyme inhibitors or angiotensin II blockers is recommended for nephroprotective purposes.

Surgical treatment. In cases of end-stage renal failure, hemodialysis and/or kidney transplantation are necessary according to standards and clinical guidelines for managing this pathology.

For patients with atrioventricular block, pacemaker implantation is performed according to applicable clinical standards and recommendations, and for life-threatening arrhythmias, implantable cardioverter-defibrillator implantation is performed, respectively [8, 24]. For skin manifestations, removal of angiokeratomas is recommended in consultation with the patient.

Conclusion

The results of the literature analysis indicate that in recent years, due to a deeper understanding of this pathology, diagnostic algorithms have been significantly improved, including genetic screening of patients' family members, and new methods of pathogenetic treatment for FD have been introduced. However, problems of late diagnosis of FD and, consequently, late initiation of specific therapy persist, which affects the prognosis and quality of life of patients. Further in-depth study of the complex issues of disease pathophysiology will allow for the improvement and enhancement of diagnostic and treatment stages for this pathology and, accordingly, optimize the individualized approach to managing this category of patients in real clinical practice.

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