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Case Report

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A Case report on Fabry disease

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Abstract

Background: Fabry disease is a rare X-linked lysosomal storage disorder caused by pathogenic variants in the GLA gene, resulting in deficient or absent α-galactosidase A enzyme activity. Progressive accumulation of globotriaosylceramide (Gb3) within various tissues leads to multisystem involvement, including the kidneys, heart, peripheral nervous system, skin, and eyes. Because the early clinical manifestations are nonspecific, the disease is frequently misdiagnosed, resulting in delayed treatment and irreversible organ damage.

Case Presentation: A 32-year-old male presented with a 15-year history of recurrent burning pain in both hands and feet, heat intolerance, hypohidrosis, recurrent abdominal pain, fatigue, and reduced exercise tolerance. Physical examination revealed angiokeratomacorporisdiffusum and bilateral corneal verticillata. Laboratory investigations demonstrated persistent proteinuria and mild renal impairment, while echocardiography showed concentric left ventricular hypertrophy. Leukocyte α-galactosidase A enzyme activity was markedly reduced, plasma lyso-Gb3 levels were significantly elevated, and molecular genetic testing identified a pathogenic GLA gene mutation, confirming classical Fabry disease. The patient was treated with enzyme replacement therapy using agalsidase beta (1 mg/kg intravenously every two weeks), along with renoprotective therapy, neuropathic pain management, and supportive care. During 12 months of follow-up, the patient experienced significant improvement in neuropathic pain, gastrointestinal symptoms, exercise tolerance, and quality of life, with stabilization of renal and cardiac function.

Conclusion: This case highlights the importance of considering Fabry disease in young patients presenting with unexplained neuropathic pain and multisystem manifestations. Early diagnosis through enzyme assay and genetic testing allows timely initiation of disease-specific therapy, which can slow disease progression, preserve organ function, and improve long-term outcomes. Greater clinical awareness and multidisciplinary management are essential to reduce diagnostic delays and optimize patient care.

Keywords: Fabry disease; Lysosomal storage disorder; α-Galactosidase A deficiency; Enzyme replacement therapy; GLA gene; Neuropathic pain.

Introduction

Fabry disease is a rare inherited metabolic disorder that belongs to a group of conditions known as lysosomal storage diseases. It occurs because of mutations in the GLA gene, which result in reduced or absent activity of the enzyme α-galactosidase A. This enzyme normally helps break down a fatty substance called globotriaosylceramide (Gb3). When the enzyme is deficient, Gb3 gradually accumulates in the cells of different organs, especially the kidneys, heart, blood vessels, skin, eyes, and nervous system. As this accumulation continues over many years, it causes progressive damage to these organs. Although Fabry disease is uncommon, it is clinically important because early diagnosis and treatment can slow disease progression, preserve organ function, and improve the patient's overall quality of life.[1,2]

One of the greatest challenges in Fabry disease is that its symptoms usually begin during childhood or adolescence but are often mild and nonspecific. Many patients initially experience burning pain in their hands and feet, reduced sweating, heat intolerance, abdominal pain, diarrhea, fatigue, or small reddish-purple skin lesions known as angiokeratomas. These symptoms can easily be mistaken for more common conditions such as peripheral neuropathy, rheumatic disorders, irritable bowel syndrome, or dermatological diseases. As a result, patients frequently consult several specialists over many years before receiving the correct diagnosis. During this delay, the disease continues to progress silently, eventually affecting major organs such as the kidneys, heart, and brain, where permanent damage may already have occurred before treatment begins.[3,4]

The diagnosis of Fabry disease requires careful clinical evaluation supported by appropriate laboratory investigations. In male patients, measurement of α-galactosidase A enzyme activity is the most important initial diagnostic test because enzyme levels are usually markedly reduced. However, enzyme testing alone may not be sufficient in female patients due to variable enzyme activity, making molecular genetic testing essential for confirmation. Additional investigations, including plasma lyso-Gb3 measurement, urine protein estimation, echocardiography, electrocardiography, ophthalmological examination, and renal assessment, help determine the extent of organ involvement. Because Fabry disease follows an X-linked inheritance pattern, family screening and genetic counseling are equally important to identify affected relatives early and initiate treatment before irreversible complications develop.[5,6]

The availability of disease-specific therapies has significantly improved the outlook for patients with Fabry disease. Enzyme replacement therapy using recombinant α-galactosidase A has become the standard treatment and helps reduce glycosphingolipid accumulation, relieve symptoms, preserve renal and cardiac function, and slow disease progression when started early. In addition to enzyme replacement therapy, patients require supportive management for neuropathic pain, kidney disease, cardiac complications, and gastrointestinal symptoms, along with regular long-term follow-up by a multidisciplinary healthcare team. This case report describes a young man with classical Fabry disease who presented with long-standing multisystem manifestations. The report highlights the importance of recognizing characteristic clinical features, confirming the diagnosis through enzyme assay and genetic testing, and initiating timely treatment to achieve favorable clinical outcomes.[7,8]

Case presentation

Patient Demographics

A 32-year-old male presented to the Department of General Medicine at a tertiary care teaching hospital with complaints of recurrent burning pain in both hands and feet, decreased sweating, progressive fatigue, recurrent abdominal pain, and reduced exercise tolerance. He was employed as a software engineer and belonged to a middle socioeconomic family. The patient reported that

his symptoms had been present since adolescence and had progressively worsened over the years. Despite multiple consultations with physicians from various specialties, including neurology, gastroenterology, and dermatology, no definitive diagnosis had been established. He was referred to the nephrology department after routine health screening revealed persistent proteinuria and mildly elevated serum creatinine levels, raising suspicion of an underlying systemic disorder.

Chief Complaints

- Recurrent burning pain in both hands and feet for 15 years
- Heat intolerance for 12 years
- Decreased sweating (hypohidrosis) for 10 years
- Recurrent abdominal pain with intermittent diarrhea for 8 years
- Progressive fatigue for 2 years
- Reduced exercise tolerance for 1 year
- Bilateral pedal edema for 3 months

History of Present Illness

The patient was apparently healthy until the age of 16 years, when he began experiencing episodes of severe burning pain involving both palms and soles. These painful episodes occurred several times each month and were aggravated by physical activity, emotional stress, fever, and exposure to hot weather. The pain often lasted for several hours to two days and was partially relieved by analgesics. Over time, the frequency and severity of these episodes increased, significantly affecting his daily activities and quality of life. He also noticed marked intolerance to heat and reduced sweating even during strenuous physical activity, making it difficult to tolerate warm environments.

Approximately eight years before presentation, the patient developed recurrent crampy abdominal pain associated with bloating, nausea, and intermittent diarrhea, particularly after meals. These symptoms were repeatedly diagnosed as irritable bowel syndrome, and treatment with antispasmodics and proton pump inhibitors provided only temporary relief. During the previous two years, he experienced progressive fatigue, generalized weakness, occasional palpitations, and reduced exercise capacity. Three months before admission, bilateral ankle swelling developed, prompting further evaluation. Routine laboratory investigations demonstrated persistent proteinuria and mildly elevated serum creatinine levels. The coexistence of neuropathic pain, gastrointestinal symptoms, renal abnormalities, and characteristic skin lesions prompted further investigations for a lysosomal storage disorder, particularly Fabry disease.

Past Medical History

The patient had no history of diabetes mellitus, hypertension, chronic kidney disease, ischemic heart disease, cerebrovascular disease, chronic liver disease, autoimmune disorders, tuberculosis, epilepsy, or malignancy. There was no history of previous major surgery or prolonged hospitalization. Since adolescence, he had received symptomatic treatment from different healthcare providers for recurrent neuropathic pain and abdominal complaints without identification of the underlying cause. No previous genetic or metabolic investigations had been performed.

Medication History

Before diagnosis, the patient had been prescribed several medications for symptomatic management, including pregabalin 75 mg once daily for neuropathic pain, diclofenac 50 mg as needed for painful episodes, dicyclomine 20 mg as needed for abdominal cramps, pantoprazole 40 mg once daily, and multivitamin supplements. He reported only partial symptom relief with these medications and admitted to irregular compliance because the symptoms frequently recurred despite treatment. He had never received enzyme replacement therapy or any disease-specific treatment before the current diagnosis.

Family History

A detailed family history revealed that the patient's maternal uncle had died from end-stage renal disease at the age of 46 years. His maternal

grandfather reportedly experienced chronic burning pain in the hands and feet and died from unexplained cardiac disease in his early fifties. The patient's mother complained of occasional burning sensations in her extremities and had recently been diagnosed with mild proteinuria but had never undergone evaluation for Fabry disease. There was no family history of diabetes mellitus, autoimmune disorders, or other inherited metabolic diseases. The pedigree suggested an X-linked pattern of inheritance.

Social History

The patient was married and lived with his family in an urban setting. He denied smoking cigarettes, alcohol consumption, or recreational drug use. His appetite was moderately reduced because of recurrent abdominal discomfort. The chronic pain and fatigue significantly affected his psychological well-being, daily functioning, and social interactions. He frequently avoided outdoor activities because of heat intolerance and inability to sweat normally. There was no history of recent travel, blood transfusions, or exposure to infectious diseases.

Occupational History

The patient had been working as a software engineer for the past ten years. His occupation involved prolonged sitting and long working hours in a temperature-controlled office environment. Although his work did not involve strenuous physical activity, periods of stress frequently triggered episodes of severe burning pain in his hands and feet. During summer months, commuting and exposure to warm temperatures often aggravated his symptoms because of impaired sweating. There was no occupational exposure to heavy metals, industrial chemicals, radiation, or environmental toxins.

Allergic History

The patient denied any history of allergy to medications, food, insect bites, or environmental allergens. There was no previous history of hypersensitivity reactions, anaphylaxis, or adverse drug reactions.

Physical Examination

General Examination

The patient was conscious, alert, cooperative, and moderately built. Mild bilateral pedal edema was present.

Vital Signs

- Temperature: 36.8°C
- Pulse rate: 86 beats/minute
- Blood pressure: 138/86 mmHg
- Respiratory rate: 18 breaths/minute
- Oxygen saturation: 98% on room air
- Body Mass Index: 24.6 kg/m²

Dermatological Examination

Multiple dark red to purple angiokeratomas were observed over the lower abdomen, groin, buttocks, and upper thighs. The skin appeared dry with markedly reduced sweating.

Ophthalmological Examination

Slit-lamp examination demonstrated bilateral corneal verticillata (corneal whorling). Visual acuity and fundus examination were normal.

Cardiovascular Examination

Normal first and second heart sounds were heard. Mild left ventricular heave was present without audible murmurs.

Respiratory Examination

Bilateral vesicular breath sounds were heard with no added sounds.

Abdominal Examination

Mild diffuse abdominal tenderness was present without hepatosplenomegaly.

Neurological Examination

Sensory examination revealed reduced pain and temperature sensation over both feet, consistent

with small-fiber neuropathy. Burning dysesthesia was present in both hands and feet, while motor power and deep tendon reflexes remained normal.

Laboratory Investigations

Complete Blood Count

Parameter	Result	Reference Range
Hemoglobin	13.8 g/dL	13–17 g/dL
Total Leukocyte Count	7,800 cells/mm³	4,000–11,000 cells/mm³
Neutrophils	62%	40–75%
Lymphocytes	30%	20–45%
Platelet Count	248,000/mm³	150,000–450,000 cells/mm³
ESR	18 mm/hr	<20 mm/hr

Renal Function Tests

Parameter	Result	Reference Range
Serum Creatinine	1.6 mg/dL	0.6–1.2 mg/dL
Blood Urea Nitrogen	32 mg/dL	7–20 mg/dL
eGFR	68 mL/min/1.73 m²	>90 mL/min/1.73 m²

Liver Function Tests

Parameter	Result
AST	28 U/L
ALT	30 U/L
Total Bilirubin	0.7 mg/dL
Serum Albumin	4.2 g/dL

Urine Examination

Parameter	Result
Protein	2+
Albumin-Creatinine Ratio	485 mg/g
Red Blood Cells	Nil
White Blood Cells	0–2/HPF
Urinary Casts	Absent

Cardiac Investigations

Electrocardiography

- Normal sinus rhythm
- Short PR interval
- Evidence of left ventricular hypertrophy

Echocardiography

- Mild concentric left ventricular hypertrophy
- Left ventricular ejection fraction: 60%
- Grade I diastolic dysfunction
- No valvular abnormalities

Neurological Investigation

Nerve conduction studies demonstrated findings suggestive of small-fiber peripheral neuropathy.

Ophthalmological Investigation

Slit-lamp examination confirmed bilateral corneal verticillata, a characteristic ocular feature of Fabry disease.

Dermatological Investigation

Skin biopsy of the angiokeratoma lesions demonstrated dilated superficial dermal capillaries with overlying hyperkeratosis, consistent with angiokeratoma corporis diffusum.

Enzyme Assay

Measurement of leukocyte α-galactosidase A enzyme activity demonstrated a markedly reduced level of 0.7 nmol/hour/mg protein (reference value >20 nmol/hour/mg protein), strongly suggestive of classical Fabry disease.

Plasma Biomarker

Plasma globotriaosylsphingosine (lyso-Gb3) concentration was markedly elevated at 42.8 ng/mL, supporting the diagnosis of Fabry disease.

Molecular Genetic Testing

Next-generation sequencing of the GLA gene identified a hemizygous pathogenic missense mutation (c.644A>G; p.Asn215Ser), confirming the diagnosis of Fabry disease.

Final Diagnosis

Based on the patient's long-standing history of recurrent burning pain in the hands and feet, reduced sweating, heat intolerance, recurrent abdominal pain, characteristic angiokeratoma skin lesions, corneal verticillata, persistent proteinuria, mild renal impairment, and evidence of left ventricular hypertrophy, Fabry disease was strongly suspected. Laboratory investigations further supported the diagnosis by demonstrating markedly reduced α-galactosidase A enzyme activity and significantly elevated plasma lyso-Gb3 levels. Definitive confirmation was achieved through molecular genetic testing, which identified a pathogenic mutation in the GLA gene. These combined clinical, biochemical, and genetic findings established the diagnosis of classical Fabry disease with multisystem involvement affecting the nervous system, kidneys, heart, skin, eyes, and gastrointestinal tract.

Differential Diagnosis

The differential diagnosis included small-fiber peripheral neuropathy, hereditary sensory and autonomic neuropathy (HSAN), diabetic peripheral neuropathy, multiple sclerosis, systemic lupus erythematosus, vasculitic neuropathy, amyloidosis, and mitochondrial disorders, as these conditions can present with neuropathic pain and multisystem involvement. Chronic kidney disease of other etiologies and hypertrophic cardiomyopathy were also considered because of the patient's proteinuria and left ventricular hypertrophy. However, the presence of angiokeratomacorporisdiffusum, hypohidrosis, corneal verticillata, markedly reduced α-galactosidaseA enzyme activity, elevated plasma lyso-Gb3 levels, and identification of a pathogenic GLA gene mutation confirmed the diagnosis of classical Fabry disease, effectively excluding the other differential diagnoses.

Treatment

Following confirmation of classical Fabry disease by markedly reduced α-galactosidaseA enzyme activity and identification of a pathogenic GLA gene mutation, disease-specific therapy was initiated with Agalsidase beta, an enzyme replacement therapy (ERT) approved for the treatment of Fabry disease. The patient received

Agalsidase beta at a dose of 1 mg/kg body weight administered intravenously once every two weeks. The enzyme preparation was diluted in 0.9% sodium chloride and infused over approximately 3–4 hours, beginning at a slow infusion rate and gradually increasing according to patient tolerance. Vital signs, including blood pressure, pulse rate, respiratory rate, temperature, and oxygen saturation, were monitored before, during, and after each infusion. Baseline and periodic assessment of renal function, cardiac function, plasma lyso-Gb3 levels, and anti-drug antibodies were planned to evaluate therapeutic response and detect potential infusion-related complications. The patient tolerated the enzyme replacement therapy well without significant infusion reactions.

To reduce the risk of infusion-associated reactions, the patient received Paracetamol 650 mg orally and Cetirizine 10 mg orally approximately 30–60 minutes before each enzyme infusion. During the initial infusion sessions, emergency medications, including Hydrocortisone 100 mg intravenously, Pheniramine maleate 22.75 mg intravenously, Adrenaline (1:1000), and oxygen therapy, were kept readily available for immediate management of hypersensitivity reactions if required. Continuous observation was maintained throughout the infusion and for one hour afterward. Since the patient remained clinically stable without fever, rash, dyspnea, hypotension, or other infusion-related adverse effects, subsequent infusions were completed without interruption.

The patient's recurrent episodes of acroparesthesia and chronic neuropathic pain were managed with Pregabalin 75 mg orally twice daily, which was gradually titrated to 150 mg twice daily according to symptom control and tolerability. Because persistent neuropathic pain continued to affect daily activities, Duloxetine 30 mg orally once daily was initiated and later increased to 60 mg once daily after four weeks, resulting in further improvement in pain intensity and overall quality of life. Paracetamol 500–650 mg orally every 6 hours as needed was advised for breakthrough pain. Nonsteroidal anti-inflammatory drugs were avoided because of the patient's renal involvement and the potential risk of worsening kidney function. The patient was also advised to avoid excessive physical exertion, dehydration, and exposure to high environmental temperatures, which are recognized triggers for Fabry pain crises.

Persistent proteinuria and mildly reduced renal function indicated early Fabry nephropathy. To reduce proteinuria and delay progression of chronic kidney disease, treatment with Ramipril 5 mg orally once daily was initiated and increased to 10 mg once daily according to blood pressure, serum creatinine, and serum potassium levels. The patient was advised to maintain adequate hydration, restrict excessive dietary sodium intake, and avoid nephrotoxic medications such as nonsteroidal anti-inflammatory drugs and aminoglycosides. Urinary albumin excretion, serum creatinine, estimated glomerular filtration rate (eGFR), and serum electrolytes were monitored every three months to evaluate renal response to therapy and adjust treatment as required.

Echocardiography demonstrated mild concentric left ventricular hypertrophy with preserved systolic function. Since the patient experienced intermittent palpitations, Metoprolol succinate 25 mg orally once daily was prescribed to control heart rate and reduce myocardial oxygen demand. Blood pressure remained adequately controlled with Ramipril, and no additional antihypertensive medication was required. Electrocardiography and transthoracic echocardiography were scheduled annually to monitor disease progression, ventricular hypertrophy, and the development of arrhythmias or heart failure. The patient was advised to avoid smoking, maintain a healthy body weight, and perform moderate physical activity as tolerated.

The patient's recurrent abdominal pain and dyspeptic symptoms were managed with Pantoprazole 40 mg orally once daily, administered 30 minutes before breakfast. During episodes of abdominal cramping, Hyoscinebutylbromide 10 mg orally three times daily as required was prescribed for symptomatic

relief. Since gastrointestinal symptoms in Fabry disease are primarily caused by autonomic dysfunction and glycosphingolipid deposition within the gastrointestinal tract, continued enzyme replacement therapy was expected to provide gradual improvement. Nutritional counseling emphasized small, frequent meals, adequate hydration, avoidance of fatty and spicy foods, and maintenance of balanced nutrition to minimize gastrointestinal discomfort.

The patient received Vitamin D3 60,000 IU orally once weekly for eight weeks, followed by 2,000 IU once daily as maintenance therapy because laboratory evaluation demonstrated vitamin D insufficiency. Psychological counseling was provided to address anxiety related to chronic illness and improve treatment adherence. The patient was referred for genetic counseling, and cascade screening of first-degree relatives was recommended using enzyme assay and molecular genetic testing. Follow-up visits were scheduled every three months to assess neuropathic pain, renal function, urinary protein excretion, plasma lyso-Gb3 concentration, cardiac status, and treatment tolerability. After twelve months of continuous enzyme replacement therapy, the patient demonstrated significant reduction in neuropathic pain episodes, improvement in exercise tolerance and gastrointestinal symptoms, stabilization of renal function with decreased proteinuria, and no evidence of disease progression or serious adverse drug reactions.

Follow-up and Outcome

The patient was followed regularly at 3, 6, and 12 months after initiation of enzyme replacement therapy. During follow-up, he reported a marked reduction in the frequency and severity of burning pain episodes, improved exercise tolerance, decreased heat intolerance, and significant relief from gastrointestinal symptoms. Laboratory investigations demonstrated stabilization of renal function with reduced proteinuria and stable serum creatinine levels. Echocardiography showed no progression of left ventricular hypertrophy, while neurological symptoms remained well controlled with supportive therapy. No significant infusion-related adverse reactions or treatment-related complications were observed. Overall, the patient experienced improved quality of life, functional status, and remained clinically stable with good adherence to long-term therapy.

Discussion

Fabry disease is a rare X-linked lysosomal storage disorder that is frequently overlooked because its early manifestations are nonspecific and resemble several common neurological, dermatological, and gastrointestinal disorders. This patient experienced recurrent burning pain in the extremities, heat intolerance, hypohidrosis, and abdominal discomfort for several years before the correct diagnosis was established. Such diagnostic delays are commonly reported and often result in irreversible renal and cardiac damage before disease-specific therapy is initiated. Clarke described Fabry disease as a complex multisystem disorder in which early symptoms are frequently misdiagnosed, emphasizing the importance of maintaining a high index of clinical suspicion. Similarly, Desnick and Banikazemi highlighted that early recognition and timely diagnosis are essential because enzyme replacement therapy is most beneficial before permanent organ damage develops. The clinical course observed in this patient closely parallels these published findings.[9,10]

The characteristic multisystem involvement observed in this patient reflects the progressive accumulation of globotriaosylceramide within vascular endothelial cells, kidneys, heart, peripheral nerves, skin, and other organs. Persistent neuropathic pain, angiokeratomas, corneal verticillata, proteinuria, and left ventricular hypertrophy collectively suggested classical Fabry disease rather than isolated neurological or renal pathology. Germain explained that glycosphingolipid accumulation gradually produces irreversible renal, cardiac, and cerebrovascular complications if treatment is delayed. Likewise, Palaiodimou and colleagues emphasized that Fabry disease requires a multidisciplinary approach because patients commonly present with simultaneous involvement of multiple organ systems. The manifestations documented in this patient are

highly consistent with the clinical spectrum described in these reviews and reinforce the importance of comprehensive clinical evaluation in individuals presenting with unexplained multisystem disease.[11,12]

Early diagnosis of Fabry disease remains a major clinical challenge because the initial symptoms are nonspecific and often mimic more common neurological or gastrointestinal disorders. This patient experienced recurrent acroparesthesia, heat intolerance, hypohidrosis, and abdominal pain for several years before the diagnosis was confirmed. Such delays are frequently reported and contribute to progressive, irreversible organ damage involving the kidneys, heart, and nervous system. Measurement of α-galactosidase A activity followed by molecular analysis of the GLA gene enabled definitive diagnosis in this patient. Germain emphasized that delayed recognition is one of the principal reasons for poor long-term outcomes in Fabry disease. Similarly, Desnick and colleagues highlighted that increased physician awareness and early biochemical testing are essential to identify affected individuals before significant organ involvement develops. These findings closely resemble the diagnostic journey observed in this patient.[13,14]

The cornerstone of Fabry disease management is early initiation of enzyme replacement therapy (ERT), which slows glycosphingolipid accumulation and delays progression of renal and cardiac complications. In this case, treatment with agalsidase beta (1 mg/kg every two weeks), together with supportive renoprotective and symptomatic therapy, resulted in significant improvement in neuropathic pain, stabilization of renal function, and better quality of life during follow-up. Wilcox and the International Fabry Disease Study Group demonstrated that long-term agalsidase beta therapy maintains stable renal function, reduces plasma globotriaosylceramide levels, and has a favorable safety profile. Likewise, Anderson et al. reported that prolonged ERT significantly decreases proteinuria progression and preserves renal and cardiac function, particularly when treatment is initiated before irreversible fibrosis develops.

The favorable clinical response observed in this patient is consistent with these published findings and highlights the importance of early disease-specific treatment.[15,16]

Conclusion

Fabry disease is a rare inherited disorder that often goes unrecognized because its early symptoms, such as burning pain in the hands and feet, reduced sweating, abdominal discomfort, and fatigue, are similar to those of many common conditions. This case highlights how delayed diagnosis can allow the disease to progress and affect multiple organs, including the kidneys, heart, skin, and nervous system. Careful clinical assessment, supported by enzyme analysis and genetic testing, played a key role in establishing the correct diagnosis. Early initiation of enzyme replacement therapy, along with appropriate supportive treatment, helped stabilize disease progression and improve the patient's symptoms and quality of life. This case emphasizes the importance of maintaining a high level of clinical suspicion for Fabry disease in young patients with unexplained multisystem manifestations. Early recognition, timely diagnosis, and multidisciplinary management are essential to reduce long-term complications, preserve organ function, and improve overall patient outcomes.

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