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| RESEARCH ARTICLE

Systemic Congestion in a Young Patient: A Case of a Rare Infiltrative Disease

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| ABSTRACT

We report the case of a 26 year old male with no known chronic medical illness who presented to the emergency department with progressive shortness of breath and generalized body swelling over two weeks. Symptoms began gradually with reduced exercise tolerance and mild lower limb edema, followed by worsening breathlessness and abdominal distension. He had no history of chest pain, fever, recent infection, or prior cardiac or renal disease. There was no relevant family history of cardiomyopathy or inherited disorders. On arrival, he was hemodynamically stable but clinically congested with elevated jugular venous pressure, bilateral basal crackles, peripheral edema, and ascites. Point of care ultrasound demonstrated a dilated inferior vena cava with minimal respiratory variation and abdominal fluid, while chest radiography showed cardiomegaly with pulmonary congestion. Initial investigations showed mild renal impairment and elevated natriuretic peptides, supporting a congestive state, while electrocardiography revealed sinus tachycardia without ischemic changes. Given the patient's young age and severity of presentation, the findings were considered atypical for conventional heart failure, raising suspicion for an underlying cardiomyopathy, particularly an infiltrative or storage disorder. The patient was started on intravenous diuretics with fluid and salt restriction, resulting in partial clinical improvement, although significant congestion persisted. Further evaluation at a tertiary center included transthoracic echocardiography and cardiac magnetic resonance imaging, which demonstrated features consistent with an infiltrative cardiomyopathy. Subsequent metabolic and genetic testing confirmed Fabry disease as the underlying diagnosis. Enzyme assays showed reduced alpha galactosidase A activity, and genetic analysis confirmed the diagnosis. This case highlights an uncommon presentation of Fabry disease manifesting as severe systemic congestion in a young adult with no prior medical history. The presence of marked jugular venous distension, dilated inferior vena cava, cardiomegaly, and ascites provided important early clues to significant cardiac involvement. Early recognition of an atypical pattern of heart failure in a young patient was essential in prompting further evaluation for infiltrative disease. The case emphasizes the importance of considering Fabry disease in young patients presenting with unexplained congestion and cardiomyopathy. Early use of bedside ultrasound and echocardiography played a key role in identifying the severity of volume overload and guiding further investigation. Confirmation of the diagnosis allowed initiation of disease specific therapy, including consideration of enzyme replacement treatment. In conclusion, Fabry disease should be considered in the differential diagnosis of young patients presenting with severe systemic congestion, particularly when imaging and clinical findings suggest cardiomyopathy without conventional risk factors. Early diagnosis is essential to allow timely treatment and prevent progressive organ damage.

| KEYWORDS

Volume Overload, Systemic Congestion, Jugular Venous Pressure, Heart Failure, Cardiomyopathy, Infiltration, Fabry Disease

| ARTICLE INFORMATION

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Introduction

Systemic congestion is a clinical state that reflects impaired venous return and elevated filling pressures, most commonly in the setting of cardiac dysfunction. It is frequently encountered in emergency and internal medicine practice and may present with a wide range of findings, including lower limb edema, ascites, hepatomegaly, and shortness of breath. While the condition is often attributed to common causes such as heart failure or fluid overload, the underlying etiology may not always be immediately apparent, particularly in younger patients with no prior medical history. In such cases, identifying the root cause is essential, as delayed diagnosis may lead to progressive organ damage and poor outcomes [10,11].

In routine clinical practice, systemic congestion is typically associated with left or right sided heart failure. Patients often present with symptoms such as exertional dyspnea, orthopnea, abdominal distension, and fatigue. Physical examination may reveal elevated jugular venous pressure, peripheral edema, and pulmonary crackles. Although these findings may point toward cardiac dysfunction, they do not provide information about the underlying cause. As a result, clinicians must consider a broad differential diagnosis, especially when the presentation is disproportionate to the patient's age or risk profile [10,13].

In younger individuals, severe systemic congestion is uncommon and should prompt further evaluation for less typical causes. Conditions such as myocarditis, congenital heart disease, and infiltrative disorders may present in this manner. Among these, infiltrative diseases are particularly important because they often remain undiagnosed until advanced stages. These conditions are characterized by the accumulation of abnormal substances within tissues, leading to progressive organ dysfunction. Cardiac involvement may result in restrictive physiology, impaired filling, and eventually symptoms of congestion [12,14].

Fabry disease is a rare inherited metabolic disorder that belongs to the group of lysosomal storage diseases. It is caused by mutations in the GLA gene, leading to deficiency or absence of the enzyme alpha galactosidase A. This enzyme defect results in the accumulation of globotriaosylceramide within various tissues, including the heart, kidneys, and nervous system. Over time, this accumulation leads to structural and functional damage, which may manifest as multisystem disease [4,6].

The clinical presentation of Fabry disease is highly variable and often depends on the degree of enzyme deficiency and the pattern of organ involvement. Early symptoms may include neuropathic pain, heat intolerance, and gastrointestinal disturbances. However, these features are frequently nonspecific and may be overlooked or misattributed to more common conditions. As the disease progresses, more serious complications may develop, including renal impairment, cardiac involvement, and cerebrovascular events [4,14].

Cardiac involvement in Fabry disease is one of the major contributors to morbidity and mortality. The disease can affect the myocardium, conduction system, and coronary microvasculature. Over time, progressive accumulation of glycosphingolipids within cardiomyocytes leads to left ventricular hypertrophy, impaired relaxation, and eventually features of heart failure. In many cases, patients develop a form of cardiomyopathy that mimics hypertrophic or restrictive patterns, making diagnosis challenging without a high index of suspicion [2,5].

One of the key clinical challenges in Fabry disease is that cardiac manifestations may be the predominant or even the initial presentation, particularly in young adults. Patients may present with unexplained left ventricular hypertrophy, arrhythmias, or symptoms of heart failure without traditional risk factors. In such scenarios, the diagnosis is often delayed, as more common conditions are considered first. This delay can result in ongoing disease progression and irreversible organ damage [2,3].

Systemic congestion in Fabry disease is typically a consequence of advanced cardiac involvement. As diastolic dysfunction worsens, ventricular filling becomes impaired, leading to elevated filling pressures and venous congestion. Patients may develop peripheral edema, ascites, and hepatic congestion, which can resemble other forms of heart failure. However, the underlying mechanism differs, as the primary issue is related to myocardial infiltration rather than ischemic or valvular disease [5,7].

Renal involvement is another important feature of Fabry disease and may contribute to the overall clinical picture. Progressive deposition of glycosphingolipids within the kidneys leads to proteinuria and declining kidney function. In advanced stages,

patients may develop chronic kidney disease, which can further exacerbate fluid retention and systemic congestion. The coexistence of cardiac and renal involvement often complicates management and worsens prognosis [7,9].

The diagnosis of Fabry disease requires a combination of clinical suspicion, laboratory testing, and genetic confirmation. Measurement of alpha galactosidase A activity is a key initial test, particularly in male patients. In females, enzyme levels may be normal, and genetic testing is often required to confirm the diagnosis. Additional investigations such as cardiac imaging, kidney biopsy, and biomarker assessment may provide supportive evidence and help determine the extent of organ involvement [4,14].

Imaging plays a central role in the evaluation of patients with suspected cardiac involvement. Echocardiography may reveal left ventricular hypertrophy, diastolic dysfunction, and other structural abnormalities. Cardiac magnetic resonance imaging can provide further detail, including the presence of late gadolinium enhancement, which reflects myocardial fibrosis. These findings, although not specific, can raise suspicion for infiltrative or storage disorders when seen in the appropriate clinical context [2,5].

One of the reasons Fabry disease remains underdiagnosed is the overlap of its clinical features with more common conditions. For example, left ventricular hypertrophy may be attributed to hypertension, while kidney involvement may be mistaken for other forms of chronic kidney disease. Similarly, symptoms such as fatigue and shortness of breath are nonspecific and may not prompt further investigation. This highlights the importance of considering Fabry disease in patients with unexplained multisystem involvement, particularly when symptoms begin at a young age [3,6].

Early diagnosis is crucial because disease specific treatment is available and may alter the course of illness. Enzyme replacement therapy has been shown to reduce substrate accumulation and improve certain clinical outcomes. More recently, additional treatment options such as pharmacological chaperones and gene based therapies have been developed. These treatments are most effective when initiated before irreversible organ damage has occurred, further emphasizing the need for timely recognition [4,8].

The prognosis of Fabry disease largely depends on the extent of organ involvement and the timing of treatment initiation. Patients with significant cardiac or renal disease are at higher risk of complications, including heart failure, arrhythmias, and end stage kidney disease. However, with appropriate management, disease progression can be slowed, and quality of life may be improved. Regular follow up and multidisciplinary care are essential components of long term management [7,8].

Case reports have played an important role in increasing awareness of atypical presentations of Fabry disease. Several reports have described patients presenting with cardiac symptoms as the initial manifestation, often leading to delayed diagnosis. In some cases, systemic congestion or heart failure was the primary presentation, highlighting the variability of clinical features. These reports emphasize the need for clinicians to remain vigilant and consider rare diagnoses when common explanations do not fully account for the clinical picture [19,20].

The presence of severe systemic congestion in a young patient without clear risk factors should raise concern for underlying structural or infiltrative disease. While common causes such as viral myocarditis or idiopathic cardiomyopathy may still be considered, the possibility of a metabolic or genetic condition should not be overlooked. A detailed history, including family history, along with targeted investigations, can provide important clues that guide diagnosis [10,11].

In clinical practice, the evaluation of such patients often requires a stepwise approach. Initial management focuses on stabilizing the patient and relieving symptoms of congestion. At the same time, efforts should be made to identify the underlying cause through appropriate testing. This may include laboratory studies, imaging, and in some cases, tissue biopsy. Collaboration between different specialties, including cardiology, nephrology, and genetics, is often necessary to reach a definitive diagnosis [10,13].

The case presented in this report describes a young patient with severe systemic congestion as the initial manifestation of previously undiagnosed Fabry disease. This presentation illustrates the diagnostic challenges associated with rare infiltrative disorders and highlights the importance of maintaining a broad differential diagnosis. Although Fabry disease is uncommon, its impact on multiple organ systems and the availability of targeted therapy make early recognition particularly important [4,6].

In summary, systemic congestion is a common clinical problem with a wide range of underlying causes. While it is most often related to conventional forms of heart failure, uncommon conditions such as Fabry disease may present in a similar manner, particularly in younger patients. The multisystem nature of Fabry disease, combined with its variable presentation, often leads to delayed diagnosis. Increased awareness and careful clinical evaluation are essential to identify affected patients and initiate appropriate treatment. This case highlights how a seemingly typical presentation of congestion can reveal a rare and clinically significant underlying disease.

Case Presentation

A 26 year old male with no known chronic medical conditions presented to the emergency department with progressive shortness of breath and generalized body swelling over a period of two weeks. The symptoms began insidiously with reduced exercise tolerance and mild lower limb edema, which gradually worsened. He later developed abdominal distension and increasing shortness of breath, particularly on minimal exertion. There was no history of chest pain, fever, cough, or recent infection. He denied any prior cardiac or renal disease and was not on regular medications. There was no known family history of cardiomyopathy or inherited disorders.

On initial assessment, the patient appeared uncomfortable and mildly tachypneic. His vital signs showed a blood pressure within normal range, mild tachycardia, and oxygen saturation slightly reduced on room air. Physical examination revealed clear signs of systemic congestion. The jugular venous pressure was markedly elevated and visible at rest, extending above the angle of the mandible while the patient was in a semi recumbent position (Figure 1). Peripheral examination showed bilateral pitting edema extending to the knees. Chest examination revealed diffuse bilateral crackles on auscultation, more prominent at the lung bases, suggestive of pulmonary congestion. Cardiovascular examination did not reveal any significant murmurs, but heart sounds were somewhat muffled. Abdominal examination showed distension with shifting dullness, consistent with ascites.

Figure 1: grossly congested jugular veins.

Point of care ultrasound was performed at the bedside and demonstrated a markedly dilated inferior vena cava with minimal respiratory variation, indicating elevated right atrial pressure (Figure 2).

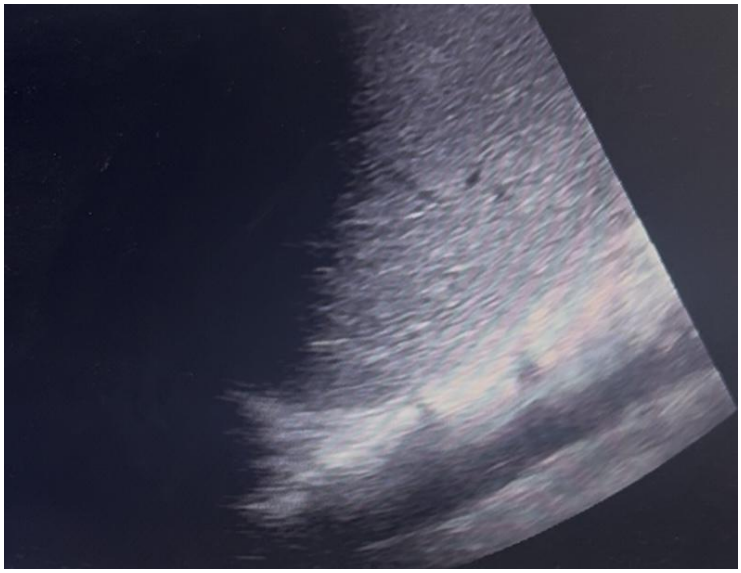

Figure 2: grossly distended inferior vena cava on ultrasonographic imaging.

Additional abdominal ultrasound confirmed the presence of free fluid within the peritoneal cavity, supporting the clinical finding of ascites (Figure 3).

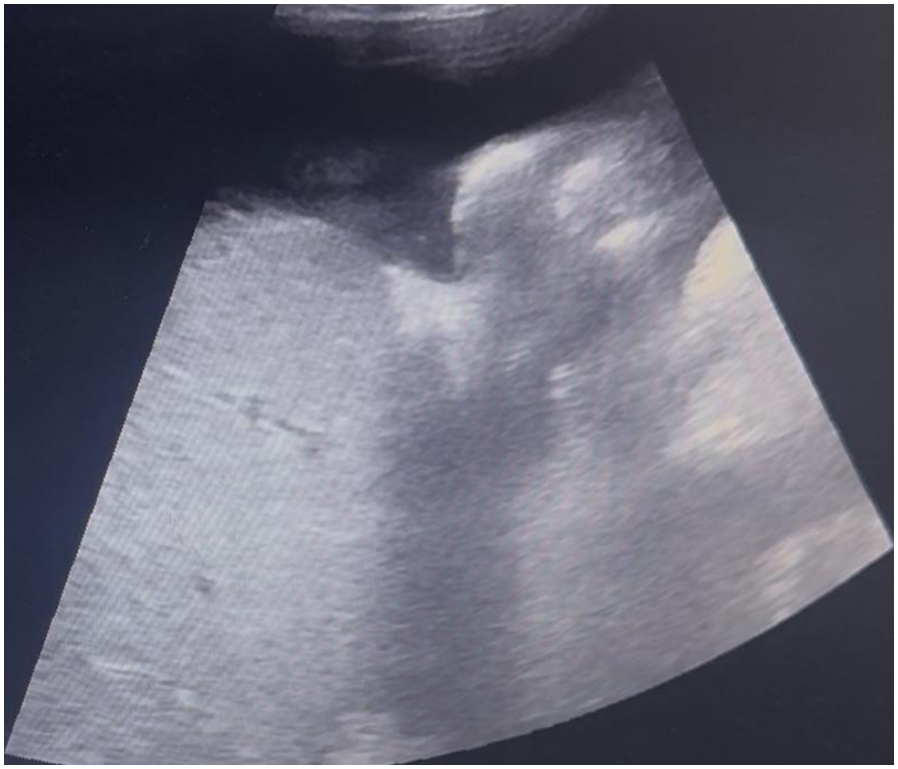

Figure 3: detection of free abdominal fluid collection in Morison's pouch, suggesting early ascites.

A chest radiograph showed cardiomegaly with prominent pulmonary vascular markings and features consistent with pulmonary congestion (Figure 4).

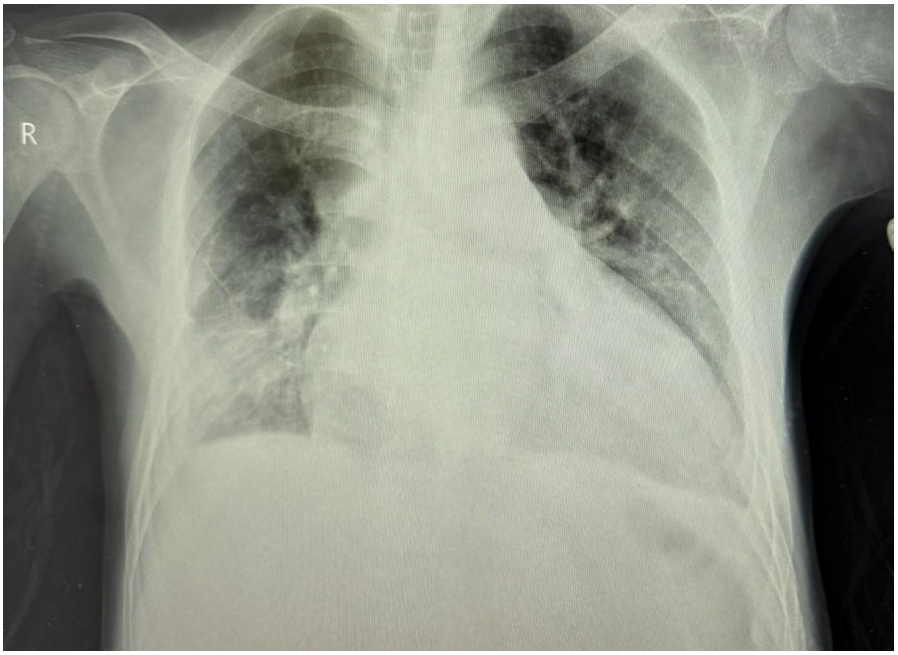

Figure 4: showing severe cardiomegaly with signs of chest congestion.

Initial laboratory investigations showed mild impairment in kidney function and elevated natriuretic peptides, consistent with a congestive state. Liver enzymes were mildly elevated, likely secondary to hepatic congestion. Cardiac enzymes were not significantly raised. Electrocardiography showed sinus tachycardia without acute ischemic changes. These findings supported the presence of significant volume overload but did not clearly identify the underlying cause.

Given the patient's young age and the severity of presentation, the clinical picture was considered atypical for conventional heart failure. The absence of known risk factors such as hypertension, diabetes, or ischemic heart disease raised concern for an alternative etiology. The combination of marked systemic congestion, cardiomegaly, and evidence of elevated filling pressures suggested the possibility of an underlying cardiomyopathy. In particular, an infiltrative or storage disorder was suspected based on the pattern of presentation.

The patient was admitted for further evaluation and management. He was started on diuretic therapy, which resulted in partial symptomatic improvement, including reduction in shortness of breath and peripheral edema. Despite this, the underlying cause of his condition remained unclear. Given the suspicion of a more complex cardiac pathology, arrangements were made to transfer the patient to a tertiary care center for advanced diagnostic workup and specialist input.

At the tertiary center, further investigations were carried out, including detailed echocardiographic assessment and advanced imaging. These studies revealed features suggestive of an infiltrative cardiomyopathy, prompting additional metabolic and genetic testing. Subsequent evaluation confirmed the diagnosis of Fabry disease as the underlying cause of his presentation.

This case highlights an unusual presentation of a rare inherited condition in a young patient with no prior medical history. The presence of marked systemic congestion at a young age, along with imaging findings such as a dilated inferior vena cava, cardiomegaly, and ascites, served as important clues to an underlying pathological process beyond typical heart failure. The use of bedside ultrasound and early imaging played a key role in identifying the severity of congestion and guiding further management.

The figures included in this report illustrate the key clinical and imaging findings observed during the initial presentation. Figure 1 demonstrates the markedly elevated jugular venous pressure on physical examination. Figure 2 shows the dilated inferior vena cava on ultrasound, reflecting increased central venous pressure. Figure 3 highlights the presence of free intraperitoneal fluid consistent with ascites. Figure 4 shows the chest radiograph with cardiomegaly and pulmonary congestion. Together, these findings provide a clear representation of the systemic nature of the patient's condition at presentation.

Management course

Management of the patient began at the time of admission when the clinical picture was consistent with significant systemic congestion. The primary concern was volume overload with evidence of elevated filling pressures, as reflected by raised jugular venous pressure, dilated inferior vena cava on ultrasound, pulmonary congestion, and ascites. Although the patient was hemodynamically stable at presentation, the severity of congestion and his young age raised concern for an underlying pathological process rather than simple fluid overload. The patient was managed initially by the internal medicine team, with early input from cardiology.

Initial management focused on symptomatic relief and stabilization. The patient was placed on continuous cardiac and oxygen saturation monitoring. Intravenous access was secured, and baseline laboratory investigations were sent. Supplemental oxygen was provided via nasal cannula to maintain adequate oxygenation. Given the degree of fluid overload, intravenous loop diuretics were initiated. A cautious dosing strategy was used, with close monitoring of urine output, renal function, and electrolytes. The patient was also placed on fluid and salt restriction to assist in reducing volume status.

Over the first 24 hours, the patient showed partial response to diuretic therapy, with increased urine output and mild improvement in shortness of breath. However, significant signs of congestion persisted, including elevated jugular venous pressure and abdominal distension. Daily clinical assessment was performed, including monitoring of body weight, fluid balance, and physical examination findings. Repeat bedside ultrasound continued to show a dilated inferior vena cava with minimal respiratory variation, indicating persistently elevated right sided pressures.

Further evaluation was undertaken to identify the underlying cause of the patient's condition. Electrocardiography and initial laboratory results did not suggest acute ischemia or infection. Given the presence of cardiomegaly on chest radiograph and ongoing congestion despite initial therapy, transthoracic echocardiography was arranged. The study revealed features suggestive of cardiomyopathy, with impaired diastolic function and findings raising suspicion for an infiltrative process. There were no significant valvular abnormalities to explain the presentation.

At this stage, the clinical suspicion for an infiltrative or metabolic cardiomyopathy increased. The patient's young age, absence of conventional risk factors, and the pattern of cardiac involvement made common causes of heart failure less likely. In view of this, and recognizing the need for advanced diagnostic testing, a decision was made to refer the patient to a tertiary care center with expertise in complex cardiomyopathies.

Prior to transfer, the patient continued to receive supportive care. Diuretic therapy was optimized, resulting in gradual reduction in peripheral edema and slight improvement in respiratory symptoms. Electrolytes were monitored regularly, and adjustments were made as needed. Renal function remained stable throughout this period. The patient remained hemodynamically stable, with no requirement for inotropic or vasopressor support.

Upon transfer to the tertiary center, a multidisciplinary team including cardiology, nephrology, and metabolic specialists became involved in the patient's care. Advanced imaging was performed, including detailed echocardiographic assessment and cardiac magnetic resonance imaging. These studies demonstrated findings consistent with an infiltrative cardiomyopathy, including increased ventricular wall thickness and abnormalities suggestive of myocardial involvement beyond typical hypertrophic patterns.

In light of these findings, further investigations were directed toward identifying a specific underlying cause. Enzyme assays were performed to evaluate for lysosomal storage disorders, and genetic testing was arranged. Additional laboratory tests were also conducted to assess for other potential causes of infiltrative disease. During this period, the patient continued to receive supportive treatment, including diuretics and careful fluid management, which led to gradual clinical improvement.

The diagnosis of Fabry disease was subsequently confirmed based on reduced alpha galactosidase A activity and genetic analysis. This finding provided a unifying explanation for the patient's cardiac and systemic manifestations. Following confirmation of the diagnosis, disease specific therapy was considered. The patient was evaluated for enzyme replacement therapy, and discussions were held regarding the benefits and long term management plan.

During his stay at the tertiary center, the patient's symptoms continued to improve. There was noticeable reduction in peripheral edema and abdominal distension, and his exercise tolerance gradually increased. Repeat imaging showed partial improvement in congestion, although underlying structural cardiac changes persisted. The patient remained stable throughout, without any acute complications.

Education and counseling formed an important part of management following diagnosis. The patient was informed about the nature of Fabry disease, its genetic basis, and the potential for multisystem involvement. Family screening was also discussed, given the inherited nature of the condition. Arrangements were made for follow up with specialized clinics for ongoing management and monitoring of cardiac, renal, and other organ involvement.

Before discharge, the patient was transitioned to an optimized medical regimen, including oral diuretics and supportive therapy as needed. Plans were made to initiate enzyme replacement therapy in the outpatient setting under specialist supervision. Clear instructions were provided regarding fluid restriction, medication adherence, and recognition of worsening symptoms.

The patient was discharged in stable condition with scheduled follow up at the tertiary center. He was advised to seek immediate medical attention if he experienced recurrence of shortness of breath, rapid weight gain, or worsening edema. Ongoing care was arranged through a multidisciplinary team to ensure comprehensive management of his condition.

This case highlights the importance of early supportive management in patients presenting with severe systemic congestion, while simultaneously pursuing the underlying diagnosis. The stepwise approach, from initial stabilization to advanced investigations and targeted therapy, played a key role in improving the patient's clinical status and establishing a definitive diagnosis.

Discussion

Systemic congestion is a common clinical problem encountered across a wide range of medical settings, most frequently associated with heart failure and fluid overload states. However, in younger patients without traditional cardiovascular risk factors, the presence of severe congestion should prompt consideration of less common etiologies. The present case highlights an important and often under recognized scenario in which a rare infiltrative disorder, Fabry disease, presents initially with features of advanced congestion. This type of presentation can pose a significant diagnostic challenge, particularly when the initial clinical picture resembles more common cardiac conditions.

Fabry disease is a rare X linked lysosomal storage disorder caused by deficiency of the enzyme alpha galactosidase A. This enzymatic defect leads to progressive accumulation of glycosphingolipids within various tissues, including the myocardium, kidneys, vascular endothelium, and nervous system. Over time, this accumulation results in structural damage and organ

dysfunction, often involving multiple systems. Although the disease may begin in childhood, diagnosis is frequently delayed until adulthood due to the nonspecific nature of early symptoms [4,14].

One of the key features of Fabry disease is its variable clinical presentation. Patients may initially report symptoms such as neuropathic pain, heat intolerance, or gastrointestinal disturbances, which are often subtle and easily overlooked. As the disease progresses, more serious complications develop, particularly involving the heart and kidneys. Cardiac involvement is a major determinant of morbidity and mortality, and in some patients, it may represent the dominant clinical manifestation [2,6].

In the context of this case, the presentation with severe systemic congestion reflects advanced cardiac involvement. Fabry related cardiomyopathy is characterized by progressive thickening of the ventricular walls, impaired relaxation, and eventual diastolic dysfunction. Unlike more common causes of heart failure, the underlying mechanism is related to intracellular accumulation of substrate rather than pressure overload or ischemia. This results in a form of restrictive physiology, where ventricular filling is impaired despite relatively preserved systolic function in the early stages [5,7].

The development of systemic congestion in such patients is primarily driven by elevated filling pressures. As diastolic dysfunction worsens, left ventricular compliance decreases, leading to increased left atrial and pulmonary venous pressures. Over time, right sided pressures also rise, resulting in elevated jugular venous pressure, peripheral edema, ascites, and hepatic congestion. These findings were clearly demonstrated in the present case, with clinical and imaging evidence supporting significant volume overload [5,7].

An important aspect highlighted by this case is the age of presentation. Severe congestion in a 26 year old patient is highly unusual in the absence of congenital or acquired structural heart disease. This should prompt clinicians to consider alternative diagnoses, including infiltrative and metabolic disorders. Fabry disease, although rare, is an important consideration in this context, particularly when cardiac findings are disproportionate to the patient's age and risk profile [3,6].

The diagnosis of Fabry disease is often delayed, with studies showing that patients may experience symptoms for several years before a definitive diagnosis is made. This delay is largely due to the nonspecific nature of early manifestations and the overlap with more common conditions. In cases where cardiac involvement predominates, patients may initially be misdiagnosed with hypertrophic cardiomyopathy or other forms of non ischemic cardiomyopathy. This highlights the importance of maintaining a high index of suspicion, especially in younger patients with unexplained cardiac findings [3,4].

Imaging plays a crucial role in raising suspicion for infiltrative cardiomyopathy. Echocardiography is often the first line investigation and may reveal left ventricular hypertrophy, diastolic dysfunction, and other structural abnormalities. However, these findings are not specific to Fabry disease. Cardiac magnetic resonance imaging can provide additional information, including the presence of myocardial fibrosis, which is commonly seen in advanced disease. In the appropriate clinical context, these findings should prompt further evaluation for underlying storage disorders [2,5].

Renal involvement is another key feature of Fabry disease and often contributes to the overall clinical picture. Progressive accumulation of glycosphingolipids within the kidneys leads to proteinuria and declining kidney function. In advanced stages, reduced kidney function may contribute to fluid retention, further exacerbating systemic congestion. The coexistence of cardiac and renal involvement, as seen in many patients with Fabry disease, is associated with worse clinical outcomes [7,9].

The confirmation of Fabry disease requires specific diagnostic testing. Measurement of alpha galactosidase A activity is typically used as an initial screening test, particularly in male patients. Genetic testing is essential for confirming the diagnosis and identifying the specific mutation. Early identification is important not only for the patient but also for family members, as the disease is inherited and may affect other relatives [4,14].

Treatment options for Fabry disease have expanded significantly over recent years. Enzyme replacement therapy remains the cornerstone of management and has been shown to reduce substrate accumulation and stabilize organ function in many patients. More recently, additional therapies such as pharmacological chaperones and gene based treatments have been introduced, offering further options for selected patients. These therapies are most effective when initiated early, before irreversible organ damage has occurred [4,8].

The present case also underscores the importance of a multidisciplinary approach in managing complex patients. The initial presentation required stabilization and management of congestion, while the subsequent diagnostic process involved advanced imaging and specialized testing. Collaboration between general physicians, cardiologists, and metabolic specialists was essential in reaching a definitive diagnosis and initiating appropriate treatment. This approach is strongly supported in the literature as a key factor in improving outcomes in rare diseases [10,11].

Case reports describing atypical presentations of Fabry disease are valuable in increasing clinical awareness. Several reports have documented patients presenting with cardiac manifestations as the first indication of disease, including heart failure, arrhythmias, and unexplained left ventricular hypertrophy. In some cases, systemic congestion was a prominent feature, similar to the presentation described here. These reports highlight the need to consider Fabry disease in the differential diagnosis of unexplained cardiomyopathy, particularly in younger patients [19,20].

Another important consideration is the potential for misdiagnosis. Given the overlap in clinical and imaging findings, patients with Fabry disease may initially be treated for more common conditions such as hypertensive heart disease or idiopathic cardiomyopathy. This can lead to delays in appropriate treatment and continued disease progression. Increased awareness and targeted screening in selected populations may help reduce diagnostic delays and improve patient outcomes [3,6].

From a practical standpoint, this case emphasizes several key clinical lessons. First, severe systemic congestion in a young patient should not be assumed to be due to conventional heart failure without further evaluation. Second, the presence of cardiomegaly and signs of elevated filling pressures should prompt consideration of underlying cardiomyopathy. Third, when standard investigations do not provide a clear explanation, clinicians should consider rare causes, including infiltrative and metabolic disorders.

The role of bedside ultrasound is also worth emphasizing. In this case, early use of point of care ultrasound helped identify a dilated inferior vena cava and the presence of ascites, supporting the diagnosis of significant congestion. This tool is increasingly recognized as an important component of clinical assessment, allowing rapid evaluation of volume status and guiding management decisions in real time [10].

The long term prognosis of Fabry disease depends on the extent of organ involvement and the timing of treatment initiation. Patients with advanced cardiac and renal disease are at higher risk of complications, including heart failure and arrhythmias. However, with early diagnosis and appropriate therapy, disease progression can be slowed, and quality of life improved. This further highlights the importance of recognizing atypical presentations and initiating timely investigation [7,8].

In summary, this case illustrates a rare but clinically significant presentation of Fabry disease as severe systemic congestion in a young patient. It highlights the diagnostic challenges associated with infiltrative cardiomyopathies and underscores the importance of maintaining a broad differential diagnosis. Early recognition, appropriate use of imaging, and timely referral to specialized centers are essential steps in establishing the diagnosis. Increased awareness of such presentations can help clinicians identify similar cases and improve patient outcomes through earlier intervention and targeted therapy.

Conclusion

This case highlights several important clinical points in the evaluation of systemic congestion, particularly in young patients. Although congestion is commonly attributed to heart failure, its presence in a previously healthy 26 year old should prompt careful reassessment and consideration of less common causes. Clinical findings such as elevated jugular venous pressure, diffuse crackles, ascites, and imaging evidence of cardiomegaly and a dilated inferior vena cava provide clear indicators of significant volume overload, but they do not identify the underlying etiology. Early use of bedside ultrasound and chest imaging plays a key role in confirming the presence and extent of congestion and guiding initial management.

The case also emphasizes that infiltrative and metabolic cardiomyopathies, although rare, can present with advanced features of heart failure. Fabry disease in particular may remain undiagnosed until adulthood, especially when early symptoms are subtle or nonspecific. When cardiac involvement predominates, patients may present with features that closely resemble more common forms of cardiomyopathy, leading to potential delay in diagnosis. Maintaining a broad differential diagnosis is therefore essential, especially when the clinical picture is not fully explained by typical risk factors.

Initial management should focus on stabilization and relief of congestion through appropriate use of diuretics and supportive care. However, equal importance should be given to identifying the underlying cause. Early referral to a tertiary center for advanced imaging and specialized testing can be crucial in such cases, as it allows for timely diagnosis and initiation of disease specific therapy. A multidisciplinary approach involving general physicians, cardiologists, and metabolic specialists is essential to ensure comprehensive evaluation and optimal patient care.

Finally, this case demonstrates how a common clinical presentation can uncover a rare but significant underlying condition. Recognition of atypical features, appropriate use of diagnostic tools, and timely escalation of care all contribute to improved patient outcomes. Increased awareness of such presentations may help clinicians identify similar cases earlier and initiate targeted management before irreversible organ damage occurs.

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