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A Woman with Loeys-Dietz Syndrome: A Case Report

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Case Report

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Abstract

Loeys-Dietz-syndrome (LDS) is a genetic connective tissue disorder, where the vascular system is primarily affected. The disease is rare, aggressive and progressive. The clinical manifestations are variable and therefore the treatment is challenging. Aortic dissection is one of the major vascular complications of LDS 1. Here, we present a case of a 60-year-old woman, lately diagnosed with LDS type 2. Until the present moment she has successfully undergone five operations for aortic aneurysms, and she has currently manifested an Arteria vertebralis aneurysm. The patient suffers from chronic leg pain and abdominal aneurysm.

Keywords: Loeys-Dietz Syndrome, Aortic Aneurysm, Dissection, Arteria Vertebralis Aneurysm, Genetic Test, Rare Disease

Introduction

Loeys-Dietz Syndrome (LDS)(1) is a rare inherited autosomal dominant progressive condition characterized mainly by vascular disorders in addition to skeletal, cutaneous and craniofacial manifestations. The condition is due to mutations in genes encoding components of the transforming growth factor-β (TGF-β) signaling pathway. Among these, TGFBR2 mutations are the most frequently identified, reported in 55-60% of cases. Molecular genetic testing is therefore of paramount importance to diagnose LDS. Given the potentially severe manifestations of LDS, early diagnosis, appropriate surveillance, and timely intervention are essential.

The vascular manifestations of LDS are the most clinically significant, with aortic dilatation and aneurysm formation representing the major determinants of morbidity and mortality. The aortic root is particularly vulnerable to dissection. Notably, disease expression can vary widely, even within families which carry the same pathogenic gene variant, reflecting marked clinical heterogeneity. Extravascular findings are usually the first prominent signs in young patients.

These include skeletal abnormalities such as joint hypermobility, which predisposes to early-onset osteoarthritis and musculoskeletal pain. In addition, pregnancy constitutes a significant risk factor for women with LDS, as it is associated with increased incidence of vascular dissections and uterine rupture, which may lead to maternal complications.

Case report

A 60-year-old female patient with three abortions, two children and no other significant prior medical history presented to the emergency department in 2011 with acute emesis, fatigue and chest pain. Physical examinations and ultrasonographic evaluation revealed an ascending aortic aneurysm measuring 5.3cm in diameter.

In September 2011 the patient underwent emergent surgical repair of an acute Stanford Type A aortic dissection,

and replacement of the aortic valve due to severe aortic valve insufficiency. In February 2013 a reoperation was required due to dissection of the equal aortic segment and a Jotec E-Vita No 30-stent graft was implanted successfully.

In 2014, the patient presented with abdominal pain and an ultrasonographic finding of a 7cm abdominal aortic aneurysm, the patient underwent operation for Type B Stanford aortic dissection repair. The postoperative course was complicated by the development of abdominal hematoma.

In 2022, during routine follow-up with computed tomography angiography (CTA) revealed an asymptomatic aortic root dissecting aneurysm measuring 57mm x 50mm. The patient subsequently underwent aortic root and aortic valve replacement.

In 2023 the patient presented severe back pain. Follow-up CTA demonstrated a descending thoracic aortic aneurysm measuring 65mm in diameter. The patient had an operation with thoracic endovascular aortic repair. It's important to highlight that despite the patient's extensive history of multifocal aneurysmal and dissecting vascular disease, genetic testing had not previously considered necessary. In 2023, at the patient's own requeste, genetic testing was performed and identified the presence of TGFBR2 gene duplication.

Therefore, Loey's Dietz Syndrome diagnosis was established approximately 12 years after the initial presentation of aortic disease. The two children were also tested for the gene, and the daughter was found positive.

The family's medical history reports sudden unexplained deaths of both the mother and aunt of the patient at the age of 80 as well as an abdominal aneurysm in the patient's mother's sister.

In 2023 the patient presented to the Emergency Department with a thoracic aortic aneurysm of 65mm with endoleak and was subjected to surgical replacement of the affected aortic segment with a prosthetic graft.

At the most recent follow up, in August 2025, the head and neck CTA revealed the presence of a right Internal Carotid Artery (ICA) intracranial aneurysm measuring 9mm with an associated 5mm dissection, as well as a Right Vertebral Artery (RVA) dissection measuring 7mm. Furthermore, an abdominal aortic aneurysm of 4-5 cm, located immediately in distal to the prior aortic prosthetic graft, was identified recently. The complete aortic reconstruction is represented in Figure 1.

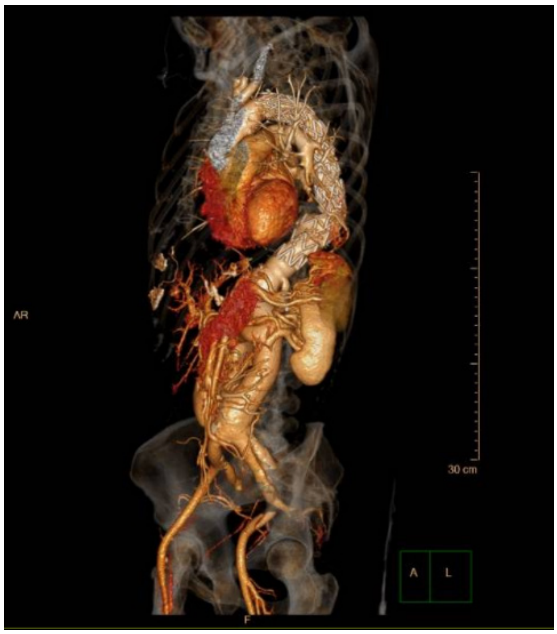

Figure 1: Angiography of the Aorta.

In 2026, the patient underwent an additional vascular intervention for an infrarenal abdominal aortic aneurysm. The procedure involved implantantion of an endovascular bifurcated prosthesis and placement of a stent graft. Follow-up imaging demonstrated stability of both intracranial ICA aneurysm and the vertebral artery dissection, with no significant interval change in size.

Currently, chronic leg pain and reduced overall physical condition pose challenges to patient's rehabilitation.

During the development of the disease the patient had fluctuating proBNP levels in blood serum, as they are demonstrated in Table 1.

DATE	proBNP VALUE	REFERENCE
2014	968.90ng/l	5-153
2017	136.70ng/l	5-153
05/2022	612 pg/ml	<125
08/2023	1226 pg/ml	<125
12/2023	513 pg/ml	<125
01/2024	542 pg/ml	<125
11/2024	490 pg/ml	<125
01/2025	366 pg/ml	<125
03/2025	565 pg/ml	<125
09/2025	401pg/ml	<125

Table 1: proBNP levels from 2014 to 2025.

Discussion

Loeys–Dietz syndrome (LDS)(5) is a rare, progressive connective tissue disorder, characterized by variable vascular and systemic manifestations. Due to its rarity (prevalence 1:50.0001) and multisystemic involvement, comprehensive and multidisciplinary coordination between health care professionals is fundamental for optimal long-term management of the patients.

The present case reports the diagnostic challenges of a patient with an initially undefined clinical entity with multiple aneurysms who was diagnosed with LDS at the age of 54 years old. This fact underlines the importance of genetic testing in LDS (7) diagnosis and highlights the significance of early detection of symptoms for the optimum management of the syndrome's complications.

Following the diagnosis, it is crucial for the patients to undergo periodic preventive checkups for large arteries manifestations given the aggressive nature of the disease5. However late in life her diagnosis was, the patient successfully underwent multiple aortic surgical and endovascular interventions and completed two pregnancies, indicating that survival is possible with the employment of right surgical management and vigilant follow-up6.

In terms of the aortic aneurysms' location, the underlying abnormality of connective tissue predisposes to progressive dilation distal to prior grafts, as it is documented in this case. Thus, in high risk of recurrent dissection individuals, total aortic arch replacement (2) - and potentially complete aortic replacement in the future - may provide long-term benefit by mitigating the risk of re-dissection and further vascular compromise near the graft 7.

Another essential issue to address is proBNP values. ProBNP serves as a nonspecific cardiac neurohormone released in response to ventricle wall stress. In literature, an increase in proBNP has been documented following surgical repair of aortic dissection, as it is also documented in this patient with elevated post-operative proBNP levels in 2014 (Figure 2).

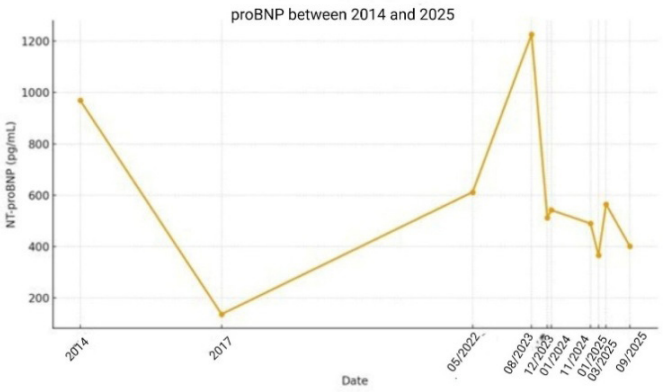

Figure 2: proBNP levels from 2014 to 2025.

Literature further associates elevated pro BNP blood serum levels with future acute aortic dissection (AAD) (3) and increased short-term mortality 3. The significantly increased pro BNP value observed in 2025 (6) may indicate ongoing aneurysmal expansion in the abdominal aorta and/or vertebral artery. Although no specific biomarker currently exists for aneurysm development, correlations between pro BNP and CRP have been described in previous studies 3. Further research is needed to evaluate pro BNP levels' potential role as early diagnostic or prognostic markers in connective tissue-related aortic pathologies.

The patient's chronic diffuse leg pain remained unexplained by neurological assessment. Therefore, it could be attributed to associated polyneuropathy, reflect abdominal aortic dissection or represent dural ectasia (4), a known feature of connective tissue disorders, that requires further imaging-based evaluation4. An intriguing aspect of this case is the intrafamilial variability of phenotypic expression, making urgent the need for family surveillance in LDS. The patient presented primarily with vascular involvement, whereas her daughter exhibits multi-system manifestations, including cataracts, skin abnormalities, bifid uvula and early-onset joint disease. This contrast may suggest an increasing phenotypic severity across generations, consistent with reports of variable expressivity in LDS1.

Finally, the psychological and social impact of LDS should not be underestimated. Repeated surgical interventions, chronic pain, reduced quality of life, constant agony about future complications and the realization of the possibility that this genetic mutation could be passed down to the next generation may cause significant emotional distress. Therefore, psychological support and family counseling should constitute an integral part of the long-term management strategy, as it has been proved to positively influence mental health and overall quality of life in chronic disorders.

In conclusion, this case underscores the progressive, multifocal, and hereditary nature of LDS and highlights the necessity for lifelong surveillance, multidisciplinary collaboration, genetic evaluation, and psychosocial support to optimize patient outcomes.

Patient consent

Written informed consent was obtained from the patient for the publication of this case report, including all anonymised clinical data and accompanying images. Documentation of consent has been retained in the patient's medical record in accordance with institutional and ethical guidelines.

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