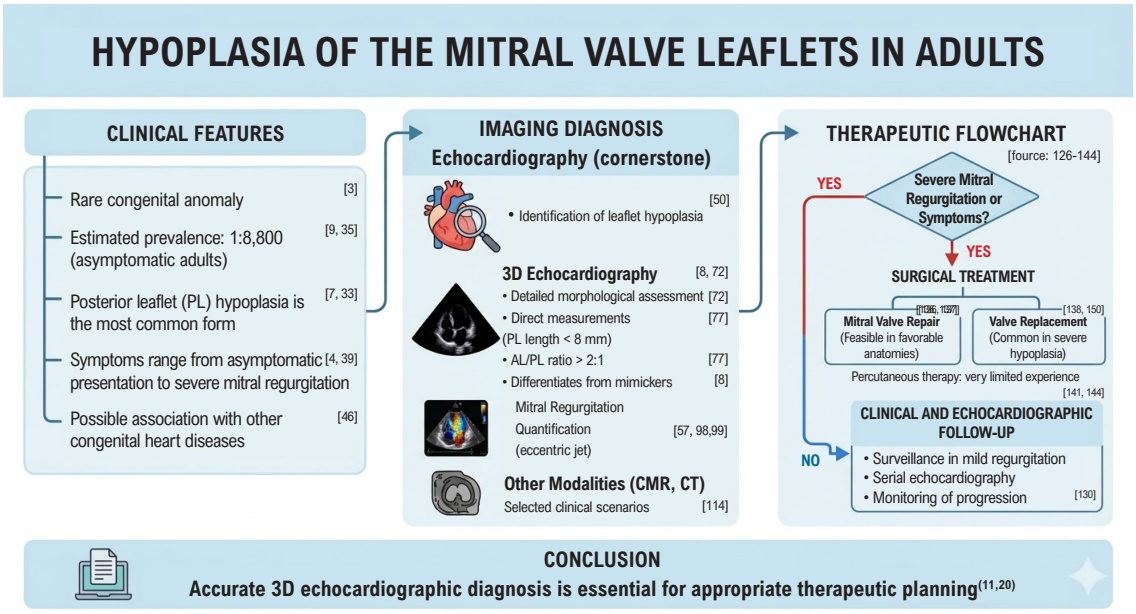
Mitral Valve Leaflet Hypoplasia in Adults: Role of Cardiovascular Imaging

Fábio Luis de Jesus Soares[1,2]
Cardio Pulmonary Hospital,[1] *Salvador, BA – Brasil*
Hospital Santa Izabel,[2] *Salvador, BA – Brasil*

Central Illustration: Mitral Valve Leaflet Hypoplasia in Adults: Role of Cardiovascular Imaging



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Mitral Valve Leaflet Hypoplasia in Adults

Abstract

Mitral valve leaflet hypoplasia is a rare congenital anomaly, traditionally described in childhood but increasingly recognized in adults, often as an incidental finding or during the evaluation of mitral regurgitation. Clinical presentation is heterogeneous and depends on leaflet anatomy, the subvalvular apparatus, and the severity of regurgitation. In this narrative literature review, including case reports, case series, and review articles from nationally and internationally recognized journals, epidemiological and clinical aspects are discussed, with particular emphasis on echocardiographic findings. Posterior leaflet hypoplasia is the most common form and may be partial or complete. Three-dimensional echocardiography plays a central role in anatomical assessment, enabling direct measurements of leaflet area and length and helping differentiate true hypoplasia from mimicking entities such as mitral cleft, functional restriction, or subvalvular abnormalities. The estimated prevalence in asymptomatic adults is approximately 1:8,800. Therapeutic management is primarily determined by the severity of mitral regurgitation, with valve repair being feasible only in selected anatomical scenarios. Therefore, refined anatomical understanding, particularly through three-dimensional echocardiography, is essential for accurate diagnosis and appropriate therapeutic planning in this rare yet clinically relevant condition.

Keywords

Mitral Valve; Congenital Abnormalities; Three-Dimensional Echocardiography.

Mailing Address: Fábio Luis de Jesus Soares •
Cardio Pulmonary Hospital. Av. Garibaldi, 2199. CEP: 40170-130. Ondina, Salvador, BA – Brazil
E-mail: fljsoares@yahoo.com.br
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Introduction

Congenital abnormalities of the mitral valve (MV) form a spectrum that includes prolapse, clefts, duplications,

congenital stenosis, subvalvular apparatus abnormalities, and, more rarely, hypoplasia of one or both leaflets. Among these, posterior leaflet hypoplasia is the most frequently described in the literature and often results in a functional unicuspid mitral valve phenotype.[1]

Historically, mitral leaflet hypoplasia was considered incompatible with life and was predominantly diagnosed in childhood in the context of severe mitral regurgitation (MR). However, over the past two decades, case reports, small series, and literature reviews have described presentations in adults, often asymptomatic or with mild symptoms, identified incidentally on routine echocardiograms[2] (Central Illustration).

From an imaging standpoint, this is a fascinating entity: the MV may maintain adequate coaptation through compensatory elongation of the opposite leaflet, annular remodeling, and left ventricular (LV) adaptations. When these mechanisms fail, MR of varying degrees predominates, generally without significant stenosis.[3]

This article reviews mitral valve leaflet hypoplasia in adults, with special focus on echocardiographic characterization, the role of other imaging modalities, and therapeutic implications.

Prevalence

Posterior mitral leaflet hypoplasia is considered a rare congenital anomaly. In a prospective analysis of 26,484 echocardiographic examinations, Bar et al. identified three cases of asymptomatic posterior leaflet hypoplasia in young adults, estimating a prevalence of approximately 1:8,800 among asymptomatic patients undergoing echocardiography.[2]

A recent systematic literature review that compiled case reports and case series identified approximately 60–70 cases of posterior leaflet hypoplasia/aplasia in adults, reinforcing the exceptional nature of the condition.[2,4] Hypoplasia of the anterior leaflet, the mitral annulus, or the entire MV (as in variants of Shone's complex) is even less frequent, with only isolated cases published.[1]

The true prevalence is likely underestimated, as mild forms without significant MR may go unrecognized on routine echocardiography, particularly when attention is focused solely on regurgitation severity rather than detailed valve morphology.

Clinical Presentation

The clinical spectrum in adulthood is broad. Reported cases range from incidental findings in asymptomatic patients, often evaluated for a soft systolic murmur, to presentations with severe MR, dyspnea, and significant left-sided chamber dilation.[5]

Reported clinical manifestations include:

- **Asymptomatic**: mild hypoplasia with preserved coaptation due to elongated anterior leaflet, without significant MR.[6]
- **Mild symptoms**: palpitations, fatigue, and exertional dyspnea, generally associated with moderate MR.[7,8]
- **Advanced presentations**: dyspnea in higher functional classes, edema, atrial fibrillation, and dilation of the left atrium and LV in the context of severe chronic MR.[3]
- In many reports, there is an **association with other congenital heart diseases**, such as: bicuspid aortic valve, ostium secundum atrial septal defect, left ventricular noncompaction cardiomyopathy, and genetic syndromes (e.g., Marfan syndrome).[4, 6]

Atypical ischemic symptoms, such as nonspecific chest pain, have also been described, although generally secondary to chronic volume overload or coexisting comorbidities rather than the hypoplasia itself.[9,10]

Echocardiographic Findings

Echocardiography is the cornerstone of the diagnosis of MV leaflet hypoplasia, allowing not only morphological identification but also hemodynamic quantification of the associated MR and evaluation of cardiac chambers.

Two-Dimensional Transthoracic Echocardiography

Typical findings include (Figure 1):

- **Marked reduction in the length of one leaflet**, most commonly the posterior leaflet, which may appear as a small rudimentary structure with limited mobility in parasternal long-axis and apical four-chamber views.[5,11]

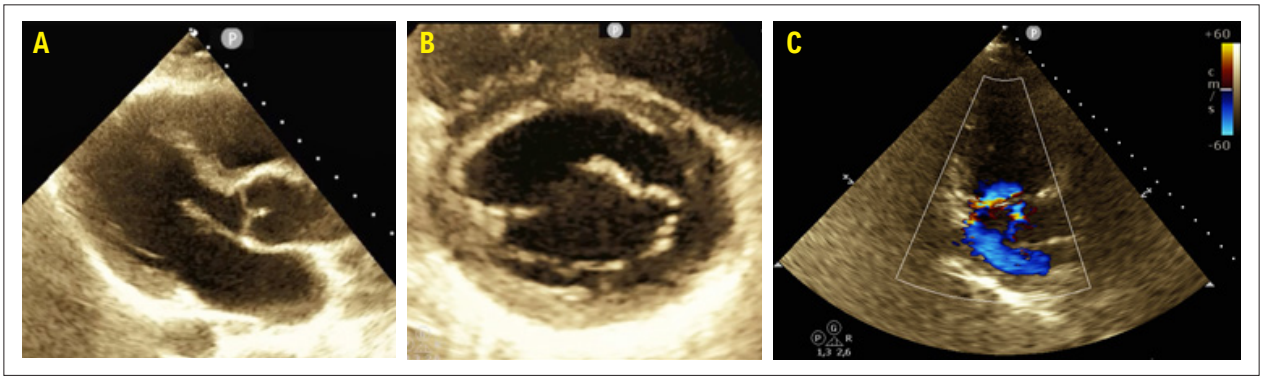

Figure 1 – *Posterior mitral leaflet hypoplasia on transthoracic echocardiography: (A) parasternal long-axis view demonstrating an anterior leaflet disproportionately elongated relative to the posterior leaflet; (B) mitral valve short-axis view demonstrating absence of stenosis; (C) apical two-chamber view demonstrating eccentric jet.*

• **Opposite leaflet (usually anterior) elongated and sometimes thickened**, projecting deeply into the ventricular cavity, often with a myxomatous appearance and occasionally associated prolapse, serving as a compensatory mechanism for coaptation.[6]

• **Coaptation line displaced** toward the hypoplastic leaflet, resulting in an eccentric MR jet directed toward that side.[4]

• **Subvalvular apparatus** generally preserved, although shortening or anomalous chordal insertion into the hypoplastic leaflet may occur, contributing to restriction.

• **Absence of significant stenosis**, with preserved valve area and low transmitral gradients in most cases; when stenosis is present, it is usually related to annular hypoplasia or more diffuse MV involvement.[12]

Some authors propose **comparative measurements**: the ratio between anterior and posterior leaflet lengths (typically > 2:1 in cases of severe posterior leaflet hypoplasia) and assessment of the effective coaptation area.[13]

Transesophageal Echocardiography (TEE) and 3D

The complex mitral anatomy requires systematic analysis, and three-dimensional echocardiography (3D TEE and 3D TTE) has become the most important tool for characterizing hypoplasia.

Although universal measurement standards have not yet been established, three parameters are consistently reported in series and case reports[3,4] (Figure 2):

a) Leaflet length

The normal posterior leaflet (PL) measures on average **10–15 mm** (varying with body surface area [BSA]). Findings suggestive of hypoplasia include:

• **Length < 8 mm** (criterion used in several case series)
• **Anterior-to-posterior leaflet ratio > 2:1**, with > 2.3–2.5:1 frequently cited in significant hypoplasia

b) Leaflet area (3D planimetry)

PL areas < **1.0–1.2 cm²** are reported in clinically relevant hypoplasia. The anterior leaflet (AL) area is generally preserved or compensatorily increased.

c) Coaptation height

Coaptation is displaced toward the hypoplastic leaflet. Abnormal findings include:

• **Coaptation height < 2 mm over PL**

• **Elongated coaptation over the AL, with displacement > 5 mm from the anatomic center**

Based on these criteria, posterior leaflet hypoplasia may be categorized as partial or total:

Partial hypoplasia

• PL present but **shortened**, **restricted,** or **underdeveloped** (Figure 3);

• Chordae often thin or abnormally inserted;

• Compensatory anterior leaflet function, maintaining some degree of coaptation.

In published cases, predominant hypoplasia includes:
• **P2**: most common (40–60% of reported cases);

• **P1**: less frequent;

• **P3**: usually associated with chordal anomalies and subvalvular restriction.

Total hypoplasia (aplasia)

Reported in a few adult cases and considered a "true unicuspid mitral valve."

• Complete anatomic absence of the PL in the atrial "en face" view (Figure 4);

• Aberrant subvalvular insertions;

• Coaptation sustained exclusively by the anterior leaflet, often markedly elongated;

• Generally associated with severe MR, although mild MR due to anterior leaflet compensation has been described.

Furthermore, 3D imaging clearly differentiates **true hypoplasia** from mitral cleft, segmental prolapse, functional restriction, and leaflet elongation without congenital hypoplasia.

Doppler and Regurgitation Quantification

Color Doppler demonstrates an eccentric jet directed opposite the hypoplastic leaflet, frequently wall-hugging (Coanda effect), which may underestimate MR severity if assessed solely by jet area (Figure 5).[3]

Therefore, the following are recommended:

• **Calculation of regurgitant volume and effective regurgitant orifice area (EROA)** using the PISA method, when feasible;

• Assessment of **vena contracta**, preferably in multiple views;

• Integration with indirect parameters: left atrial size, LV diameters and volumes, and pulmonary artery systolic pressure;

• True coaptation area;

• 3D regurgitant volume;

• Anatomic regurgitant orifice area (3D EROA), useful in eccentric jets;

• Complete mitral annular reconstruction (diameters, nonplanar angle, saddle height), frequently altered in significant hypoplasia cases.[5]

In pure hypoplasia with adequate compensation by the opposite leaflet, MR may be absent or mild; in cases with annular dilation and associated prolapse, MR tends to be severe.[13]

Other Imaging Modalities

Although echocardiography is the first-line method, **cardiac magnetic resonance (CMR)** and **computed tomography (CT)** may complement evaluation in selected situations.

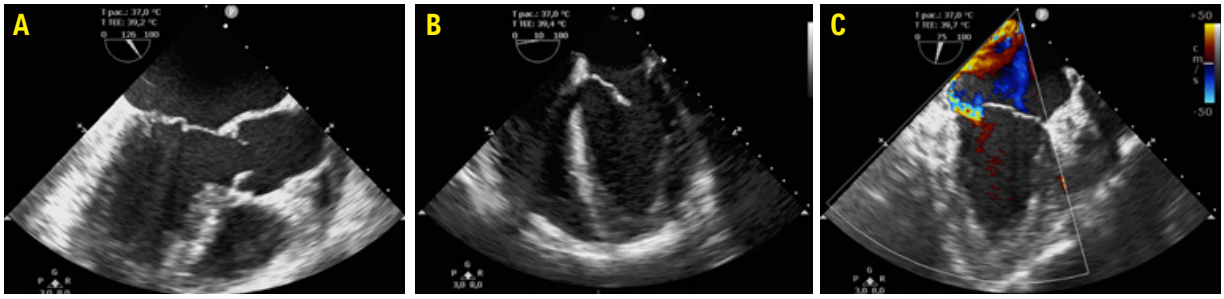

Figure 2 – *Transesophageal echocardiography demonstrating coaptation displaced toward the posterior leaflet (A), an anterior-to-posterior leaflet ratio > 2:1 (B), and the eccentric jet resulting from ineffective coaptation (C).*

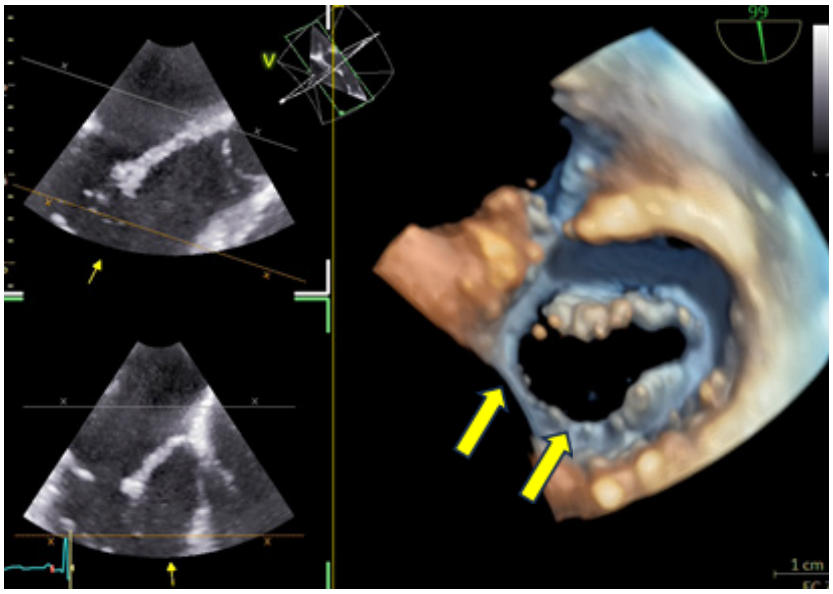

Figure 3 – *Three-dimensional transesophageal echocardiography with ventricular view of the mitral valve demonstrating partial posterior leaflet hypoplasia (segments P2 and P3 – yellow arrows).*

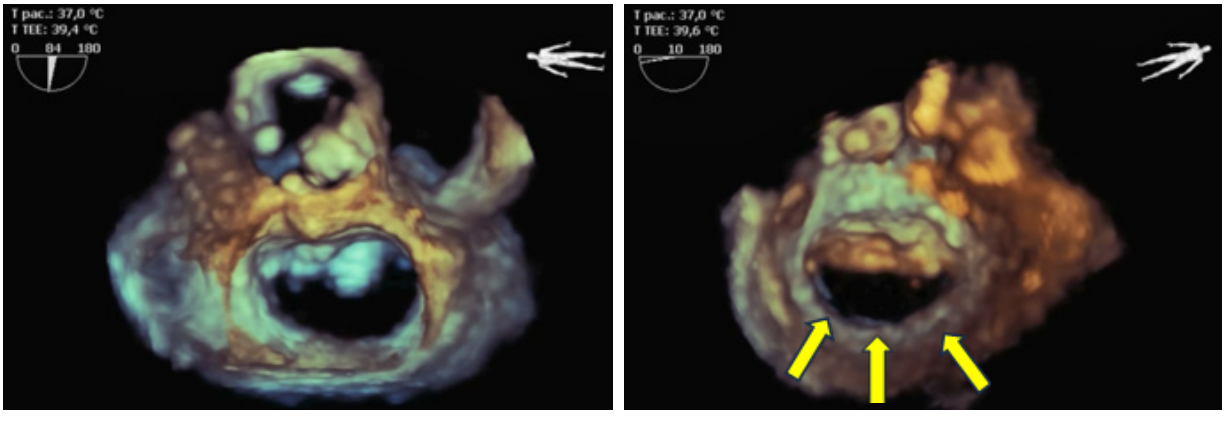

Figure 4 – *Three-dimensional transesophageal echocardiography with atrial and ventricular views of the mitral valve demonstrating complete posterior leaflet hypoplasia (segments P1, P2, and P3 – yellow arrows).*

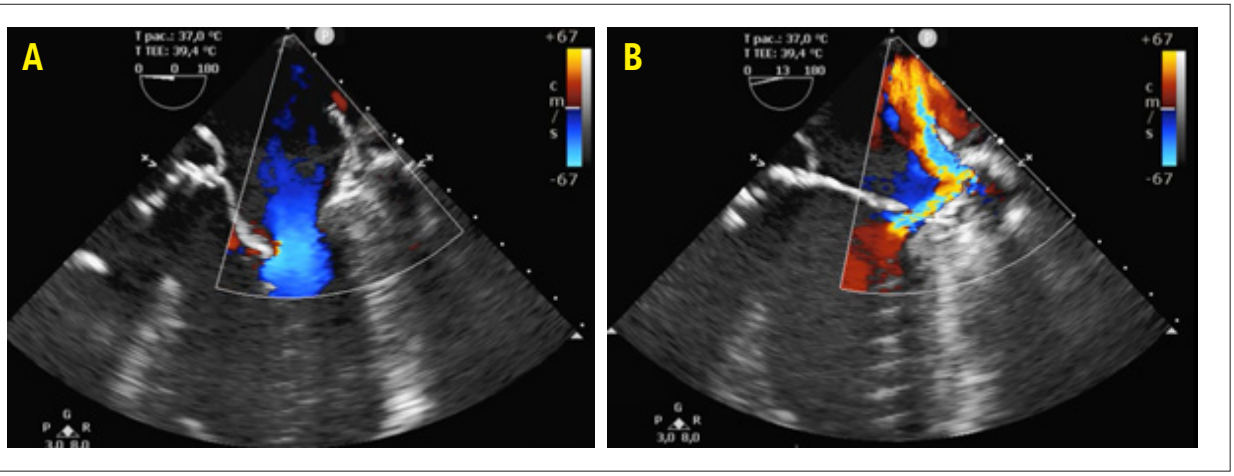

Figure 5 – *Two-dimensional transesophageal echocardiography demonstrating antegrade (A) and eccentric retrograde flows.*

Cardiac Magnetic Resonance

CMR contributes primarily in three aspects:

1. Precise volumetric and functional assessment of the LV and left atrium, useful for quantifying the impact of chronic MR and aiding surgical decision-making.

2. Regurgitation quantification by phase-contrast flow (difference between LV stroke volume and ascending aortic flow), providing a measure independent of echocardiography.

3. Tissue characterization of the mitral annulus and intervalvular fibrosa region in cases with suspected fibroelastosis, lipomatosis, or fibrosis associated with annular or anterior leaflet hypoplasia.[12]

Case reports show that CMR may confirm annular restriction with limited opening even in the absence of a significant gradient and exclude additional structural heart disease.[1]

Cardiac Computed Tomography

Cardiac CT may be useful in:

- Patients with suboptimal echocardiographic windows;
- Detailed anatomic assessment of the mitral annulus and its relationships with adjacent structures, particularly in the planning of complex surgeries or concomitant procedures (e.g., aortic valve replacement in patients with bicuspid aortic valve and mitral leaflet hypoplasia).[11]

However, due to the low prevalence of the entity and the high sensitivity of echocardiography (particularly 3D TEE), CMR and CT remain complementary rather than routine modalities.

Treatment

There are no specific guidelines for managing MV leaflet hypoplasia in adults; decisions generally follow recommendations for primary MR, adapted to the peculiar anatomic context.

Clinical and Echocardiographic Follow-up

Asymptomatic patients with mild or no MR and without significant chamber dilation may be followed clinically, with serial echocardiography to monitor MR progression, ventricular remodeling, and the emergence of symptoms or arrhythmias.[6]

Follow-up intervals are typically annual or biennial, depending on MR severity and remodeling degree.

Surgical Approach

Most adult patients described in the literature with severe MR or limiting symptoms have undergone surgical treatment. Options include:

- **Mitral valve repair**: technically challenging when the hypoplastic leaflet is very short with limited coaptation area. In some cases, leaflet augmentation with pericardium, annuloplasty, and correction of opposite leaflet prolapse are feasible;[4]
- **Mitral valve replacement:** often the most common solution in scenarios of severe hypoplasia, small annulus, or multiple associated anomalies in which durable repair is unlikely.[9]

Some reports question whether repair should always be attempted, particularly when challenging anatomy increases the risk of early reoperation.[3]

Percutaneous Therapy

Experience with **transcatheter edge-to-edge repair (MitraClip/PASCAL-type procedures)** in leaflet hypoplasia is very limited. The reduced height of the hypoplastic leaflet increases the risk of incomplete leaflet grasp, residual regurgitation, and functional stenosis after clipping.

In practice, these patients are rarely considered good candidates, except in highly selected cases with favorable opposite leaflet anatomy and high surgical risk. Current literature includes only indirect mentions, without robust series specific to this population.[3]

Conclusion

Mitral valve leaflet hypoplasia in adults, particularly of the posterior leaflet, is a rare, likely underdiagnosed entity with a

wide spectrum of clinical presentation - from incidental finding to symptomatic severe MR.

Transthoracic and transesophageal echocardiography, particularly with three-dimensional reconstruction, constitute the diagnostic cornerstone, enabling precise characterization of valve morphology, MR quantification, and planning of surgical or percutaneous interventions. Complementary modalities such as CMR and CT add anatomic and functional information in selected situations.[2,3,12]

From a therapeutic standpoint, management follows principles of primary MR, with clinical surveillance in mild cases and surgical indication in symptomatic patients or those with significant structural impact. Mitral repair may be feasible in favorable anatomies, but severe hypoplasia often leads to valve replacement. Evidence for percutaneous therapies remains limited and based on extrapolation.[2,13]

Given the still-limited number of reported cases and series, there is room for **multicenter registries** and **standardized descriptions** to better understand natural history, predictors of decompensation, and long-term outcomes of different therapeutic strategies. For the echocardiographer and cardiovascular imaging specialist, maintaining a high index of suspicion in the presence of "unusual" MV morphology is essential to avoid missing this rare - but clinically relevant - diagnosis.

Author Contributions

Conception and design of the research, data acquisition, analysis and interpretation of data, writing of the manuscript, and critical revision of the manuscript for important intellectual content: Fares FLJ.

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Study Association

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Ethics Approval and Consent to Participate

This article does not contain any studies with human participants or animals performed by any of the authors.

Use of Artificial Intelligence

The authors did not use any artificial intelligence tools in the development of this work.

Availability of Research Data

The underlying content of the research text is contained within the manuscript.

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