

Atypical Scrotal Manifestation of Madelung's Disease

Manifestação Escrotal Atípica da Doença de Madelung

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Palavras-chave: Escroto; Lipomatose Simétrica Múltipla

Madelung's disease (MD) is a rare and benign disorder of lipid metabolism, with an estimated incidence of 1:25 000. It predominantly affects middle-aged men of Mediterranean origin, typically associated with a history of chronic alcohol abuse.^{1,2}

The disease is characterized by the progressive and symmetric accumulation of non-encapsulated adipose tissue, primarily involving the proximal upper body regions. Its etiology and pathogenesis remain poorly understood.^{2,3}

Currently, there are no established diagnostic criteria. Diagnosis relies on clinical and imaging evaluation, with magnetic resonance imaging (MRI) considered the gold standard.¹⁻³

We report the case of a 64-year-old male patient diagnosed with MD, with a past medical history of alcohol-related chronic liver disease, active smoking, and a recent diagnosis of squamous cell carcinoma of the mid-esophagus (stage uT1bN1M0). He presented to the emergency department with a progressively enlarging right inguinoscrotal swelling that had been developing over three years, with significant worsening in size and pain over the preceding week.

Physical examination revealed a large bilateral scrotal mass, soft in consistency, non-tender to palpation, irreducible, and without signs of local inflammation (Fig. 1A). Given

the clinical presentation and a presumptive diagnosis of inguinal hernia, a scrotal ultrasound (US) was performed for further characterization. It revealed marked bilateral scrotal enlargement, predominantly composed of polylobulated, hyperechoic adipose tissue, without clear anatomical continuity with the inguinal canal (Fig. 1B). The imaging findings were similar to the patient's cervicofacial fat accumulation due to MD, raising the suspicion of scrotal involvement. A dedicated pelvic MRI was subsequently performed, confirming the presence of fat signal intensity tissue throughout all sequences, without suspicious contrast enhancement, consistent with scrotal involvement by MD.

Despite its typically insidious and asymptomatic progression, lipomatous masses in MD may exert compressive effects on adjacent structures. Involvement of the scrotal region is particularly relevant due to its proximity to the external genitalia, potentially leading to complications such as testicular atrophy and hidden penis syndrome, which were observed in this case.⁴⁻⁶ However, at the time of writing, the patient remains under follow-up in the General Surgery outpatient clinic, with conservative management until symptoms justify surgical intervention.

To the best of our knowledge, scrotal involvement in MD is exceedingly rare, with only a few cases described in the literature.⁶ Given this uncommon presentation, we believe this report is of particular interest, as it underscores the importance of including MD in the differential diagnosis of atypical scrotal masses – especially in chronic alcohol users with compatible phenotypic features – potentially avoiding unnecessary invasive procedures.



Figure 1 – Patient's photo of the genital area view showing marked bilateral scrotal enlargement, more prominent on the right side, causing a deformity aptly named hidden penis syndrome (A). Coronal T2-weighted MRI image of the scrotum, demonstrating large masses with high-signal intensity for fat deposition (asterisks), that causes a leftward shift of the scrotal septum (arrowhead) and superolateral displacement of the testis (arrows).

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AUTHOR CONTRIBUTIONS

PA: Writing of the manuscript.

DG, IP: Critical review of the manuscript.

All authors approved the final version to be published.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in October 2024.

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DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT


Obtained.

CONFLICTS OF INTEREST


The authors declare that they have no conflicts of interest related to this work.

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