

Doença de Madelung: A Importância da Identificação do Fenótipo Madelung's Disease: The Importance of Phenotype Identification

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Palavras-chave: Fenótipo; Lipomatose Simétrica Múltipla.

Keywords: Lipomatosis, Multiple Symmetrical; Phenotype.

Madelung's disease (MD), also known as multiple symmetric lipomatosis, is a rare lipid metabolic disorder characterized by the presence of painless, symmetric, non-encapsulated fatty deposits in the subcutaneous layer, resulting in a typical phenotype.¹⁻³ MD preferentially affects Mediterranean adult males and there appears to be an association with alcohol abuse.¹⁻³ While there are limited reports suggesting a monogenic cause, the precise etiology of the disease remains unclear.¹⁻³ Several hypotheses have been proposed, including mitochondrial dysfunction and catecholamine-induced lipolysis.¹⁻³

A 78-year-old female with a medical history of metabolic syndrome, hyperuricemia, nontoxic goitre, and a history of alcoholism, was admitted to the Internal Medicine Department with COVID-19 infection and respiratory failure. During the physical examination, the patient, who was obese, exhibited diminished breath sounds in both lungs. Her appearance was dysmorphic, displaying a muscular build with limited and protruding masses symmetrically distributed on the body surface, primarily in the neck, shoulder girdle, proximal upper limbs and trunk (Fig. 1). A computed tomography (CT) scan revealed symmetric lobulated subcutaneous fatty deposition in a centripetal distribution (Fig. 2). The thyroid gland showed only one 17 mm nodule. All laboratory results, including thyroid hormones, were normal. Based on these findings, the previous diagnosis of goitre was considered incorrect, and a diagnosis of Madelung's disease was made. ■

Declaração de Contribuição

IMC, HR – Redação do manuscrito, seleção das imagens, revisão bibliográfica, revisão crítica do conteúdo e aprovação da versão final do artigo.

IF – Revisão crítica do conteúdo e aprovação final do artigo.

Todos os autores aprovaram a versão final a ser publicada

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<https://doi.org/10.60591/crspmi.104>



Figure 1: Picture of the patient suffering from Madelung's disease type Ic, affecting the neck, shoulder girdle, upper arms and trunk.

Contributorship Statement

IMC, HR - Writing the manuscript, selecting the images, reviewing the literature, critically reviewing the content and approving the final version of the article.

IF - Critical revision of the content and final approval of the article.

All authors approved the final draft

Responsabilidades Éticas

Conflitos de Interesse: Os autores declaram a inexistência de conflitos de interesse na realização do presente trabalho.

Fontes de Financiamento: Não existiram fontes externas de financiamento para a realização deste artigo.

Confidencialidade dos Dados: Os autores declaram ter seguido os protocolos da sua instituição acerca da publicação dos dados de doentes.

Consentimento: Consentimento do doente para publicação obtido.

Proveniência e Revisão por Pares: Não comissionado; revisão externa por pares.

Ethical Disclosures

Conflicts of interest: The authors have no conflicts of interest to declare.

Financing Support: This work has not received any contribution, grant or scholarship

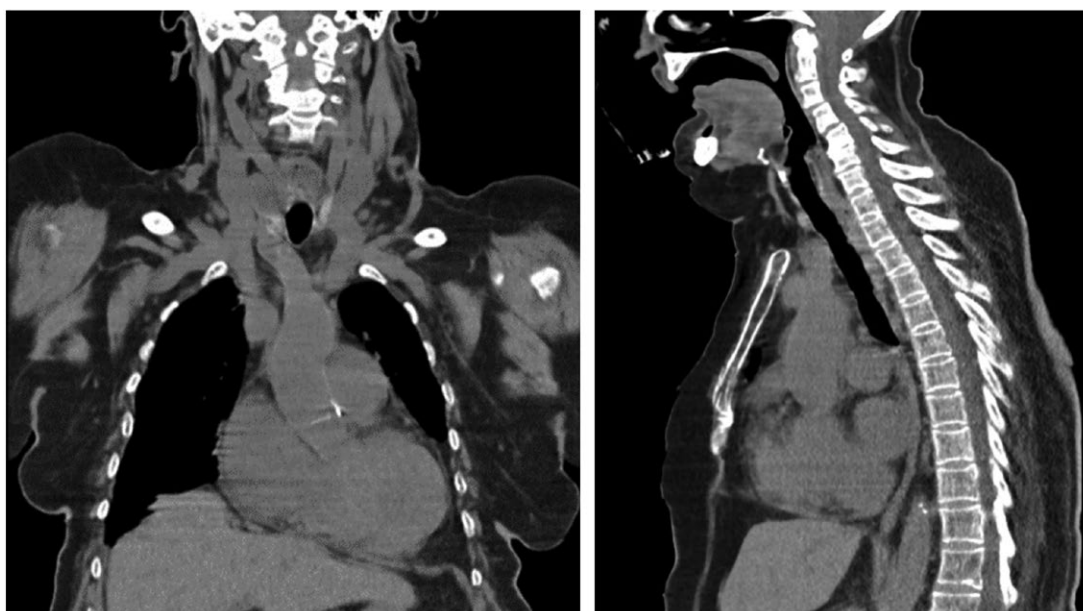


Figure 2: Cervical thoracic computed tomography (coronal and sagittal view), showing fat deposits in the neck, scapular girdle and thoracic region.

Confidentiality of Data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.
Patient Consent: Consent for publication was obtained.

Recebido / Received: 2023/08/08

Aceite / Accepted: 2023/09/20

Publicado online / Published online: 2024/07/05

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