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OPIS PRZYPADKU
CASE REPORT

A rare case of Madelung disease in a 56-year-old man: A diagnostic dilemma

Rzadki przypadek choroby Madelunga u 56-letniego mężczyzny – dylemat diagnostyczny

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ABSTRACT

Madelung disease (MD) is a rare condition characterized by numerous subcutaneous lipomas, also known in the literature as multiple symmetric lipomatosis, benign symmetric adenolipomatosis, or Launois–Bensaude syndrome. MD is little known among physicians, which makes its diagnosis difficult despite the characteristic phenotype of people affected by the disease. Although the treatment options are limited to surgical removal, prompt diagnosis and management improve patients' quality of life. The purpose of this paper is to present the case report of a 56-year-old patient admitted to our clinic for elective diagnostics of nodular subcutaneous changes in the head, neck, chest, and abdomen. During routine diagnostics, malignancy was ruled out and MD was diagnosed.

KEYWORDS

Madelung disease, multiple symmetrical lipomatosis, benign symmetric lipomatosis

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STRESZCZENIE

Choroba Madelunga (*Madelung disease* – MD) to rzadkie schorzenie, charakteryzujące się występowaniem licznych podskórnych tłuszczaków, w literaturze znane również jako mnoga symetryczna tłuszczakowatość, łagodna symetryczna adenolipomatoza lub zespół Launois i Bensaude'a. MD jest mało znana przez lekarzy, co utrudnia jej rozpoznanie pomimo charakterystycznego fenotypu osób dotkniętych chorobą. Metody leczenia ograniczają się głównie do chirurgicznego usuwania zmian, niemniej szybka diagnoza i odpowiednie postępowanie mogą znacznie poprawić jakość życia pacjentów. Celem niniejszej pracy jest przedstawienie opisu przypadku 56-letniego mężczyzny, przyjętego do kliniki w celu planowej diagnostyki guzowatych zmian podskórnych w obrębie głowy, szyi, klatki piersiowej oraz jamy brzusznej. W trakcie rutynowej diagnostyki wykluczono proces nowotworowy i rozpoznano MD.

SŁOWA KLUCZOWE

choroba Madelunga, mnoga symetryczna tłuszczakowatość, łagodna symetryczna lipomatoza

INTRODUCTION

Madelung disease (MD), also known as multiple symmetrical lipomatosis, is a rare condition characterized by the progressive development of symmetrical, benign fatty tumors that predominantly affect the upper body. These lipomas are commonly located around the neck, shoulders, upper arms, and trunk. The disease is most frequently observed in middle-aged men, often with a history of chronic alcohol consumption, althoughugh the etiopathogenesis remains poorly understood [1]. Additionally, MD is usually associated with metabolic disturbances, including insulin resistance, dyslipidemia, and an increased risk of cardiovascular diseases [2]. This condition is of particular interest because of its distinctive clinical presentation, potential complications, and the challenges involved in managing it.

In 2021, a systematic review was presented to summarize the information collected so far on MD. It was found that the highest incidence is among people aged 45–65 years, with men being significantly more likely to be affected. Most of the cases described in the literature so far come from European countries, mostly Italy and Portugal [3].

Although the clinical picture of MD is quite characteristic, observations from clinical practice indicate that its diagnosis is often difficult, suggesting the need to improve awareness of this problem among physicians. The purpose of this paper is to present the case report of a 56-year-old male patient who suffered from multiple lipomas around the head, neck, chest, and abdomen, which constituted MD.

CASE REPORT

Clinical picture and anthropometric measurements

A 56-year-old patient was admitted to our clinic for elective diagnostics of subcutaneous nodules in the head, neck, chest, and abdomen (Figures 1a,b). The symptoms began about 2 years before hospitalization and the nodules gradually increased in size over that time. The patient complained about discomfort and shortness of breath while lying down – probably due to pressure from changes in the neck area – spinal pain caused by the weight of the abdominal overhang, and difficulty moving. He declared having no chest pain, abdominal pain, constipation, diarrhea, blood in the stool, weight loss, fever, night sweats, dysphagia, or odynophagia. Also, he had no chronic diseases and had not been taking any regular medications. The patient used to smoke cigarettes but had quit about 18 months earlier. The patient declared he did not abuse alcohol. The patient reported no prior surgical procedures and no previous diagnoses related to his primary health concern. Unfortunately, the patient's family history of MD is unknown.

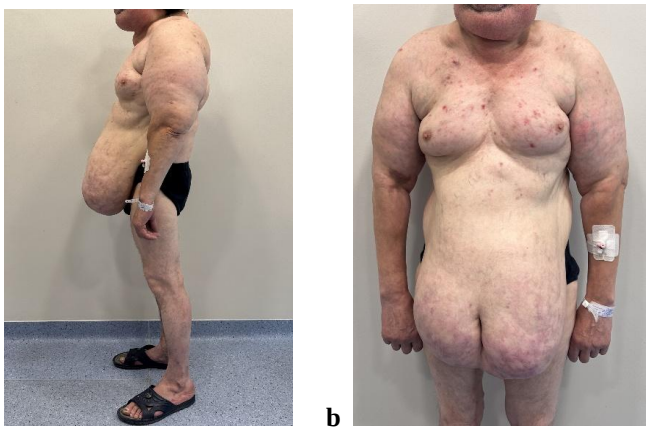

Fig. 1a,b. Multiple subcutaneous nodules in the submandibular area, the neck, and the chest visible on physical examination

Upon physical examination, attention was drawn to multiple subcutaneous nodules in the submandibular area, the neck, and the chest. They were soft, painless, and movable relative to the underlying tissue. On auscultation over the lung fields, the vesicular breath sounds were symmetric and the heart rate was regular (~80 beats per minute). On palpation, the abdomen was soft, not tender, and without internal organ enlargement. The lower extremities were symmetric, with no edema. The patient's body mass and height were 83.8 kg and 166 cm, respectively, resulting in a body mass index of 30.4 kg/m^2. The patient's body composition was analyzed using the bioelectrical impedance method performed with a TANITA MC-780, revealing that body fat accounted for 26.9 kg (32.1% of the patient's body weight).

Laboratory tests

A complete blood count showed that the red blood cell count (3.57 × 10^3/μL), hemoglobin concentration (12.3 g/dL), and hematocrit level (35.9%) were slightly decreased and mean cell volume was elevated (100.6 fL). Other parameters were all within the normal ranges, particularly the counts of other blood cells (white blood cells = 4.9 × 10^3/μL; neutrophils = 3.0 × 10^3/μL; platelet count = 172 × 10^3/μL). The serum electrolyte levels, including sodium, potassium, phosphorus, magnesium, and calcium, were within the normal ranges. The parameters for iron metabolism, i.e., iron serum concentration and total iron binding capacity, were also within the normal ranges. The serum creatinine level was 0.48 mg/dL. Gamma-glutamyl transpeptidase concentration was elevated (151.0 U/L). However, there were no aberrations in the activity of other liver and pancreatic enzymes, including alanine aminotransferase (22.5 U/L), aspartate aminotransferase (40.3 U/L), amylase (62.0 U/L), and alkaline phosphatase (69.0 U/L); the total bilirubin serum concentration was normal (0.350 mg/dL). No features of myocardial injury were found, assessed by cardiac troponin T serum concentration (6.1 pg/mL). No features of heart failure were found, as the concentration of prohormone of brain natriuretic peptide was normal (19.4 pg/mL). The coagulation parameters were normal (activated partial thromboplastin = time 26 s; prothrombin time = 10.8 s; international normalized ratio = 1.1). The D-dimer concentration in the blood was elevated (1.029 mg/L). Fibrinogen serum concentration was normal (2.1 g/L). No features of acute inflammation were found, as assessed by C-reactive protein serum concentration (2.82 mg/L), although the erythrocyte sedimentation rate was slightly high (19 mm/h). Both fasting glucose and glycated hemoglobin levels were normal (85.8 mg/dL and 5.19%, respectively). All lipid fractions, thyroid hormones, and thyroid stimulating hormone were within normal ranges. The general urine test did not show any deviations from the norm, either in physical parameters or sediment.

Diagnostic imaging and endoscopy

Abdominal ultrasound showed an enlarged liver with significantly increased echogenicity. The gallbladder, common bile duct, and intrahepatic bile ducts were normal. The portal vein and abdominal aorta were not dilated, although a few atherosclerotic plaques in abdominal part of aorta were noted. The spleen and both kidneys were in the normal position and had normal size and structure, but in the right kidney, there was a cortical cyst measuring 11.1 mm. The urinary bladder was well filled with smooth walls and the prostate was homogeneous and not enlarged. No free fluid was observed in the peritoneal cavity. Due to the multiple subcutaneous nodules in the head, neck, chest, and abdomen, computed tomography (CT) with contrast was performed on the affected areas. In the CT of the abdomen and minor pelvis, there was a large amount of fat tissue with slightly increased density in the subcutaneous layers along the anterior surface of the body, forming a protrusion with maximal dimensions of about 400 × 400 × 100 mm, starting from the level of the xiphoid process of the sternum and gradually increasing downwards (Figure 2). Imaging of the other abdominal organs did not show any significant pathological changes. The CT scan of the chest showed chronic inflammatory and post-inflammatory nodules in both lungs and a large amount of fat tissue in the body layers. Moreover, degenerative and proliferative changes of the spine were described. The CT of the neck revealed a significantly thickened fatty tissue layer, up to 50 mm below the occipital bone (Figure 3), about 75 mm at the level of the neck, about 55 mm forward from the hyoid bone (Figure 4), and about 45 mm at the level of the upper chest, with fibrous septa modulating the surrounding tissue structures. Hypertrophy of the fatty tissue was also visible within the neck muscles. No areas of pathological contrast enhancement were observed. A thyroid ultrasound was performed, showing slightly heterogeneous echogenicity of the thyroid without organ enlargement. The examination was difficult due to a thick fat layer in the area. Due to mild macrocytic anemia, endoscopic examination was performed. Gastroscopy revealed mild gastritis in the area of the gastric antrum, a healed ulcer, and *Helicobacter pylori* infection.

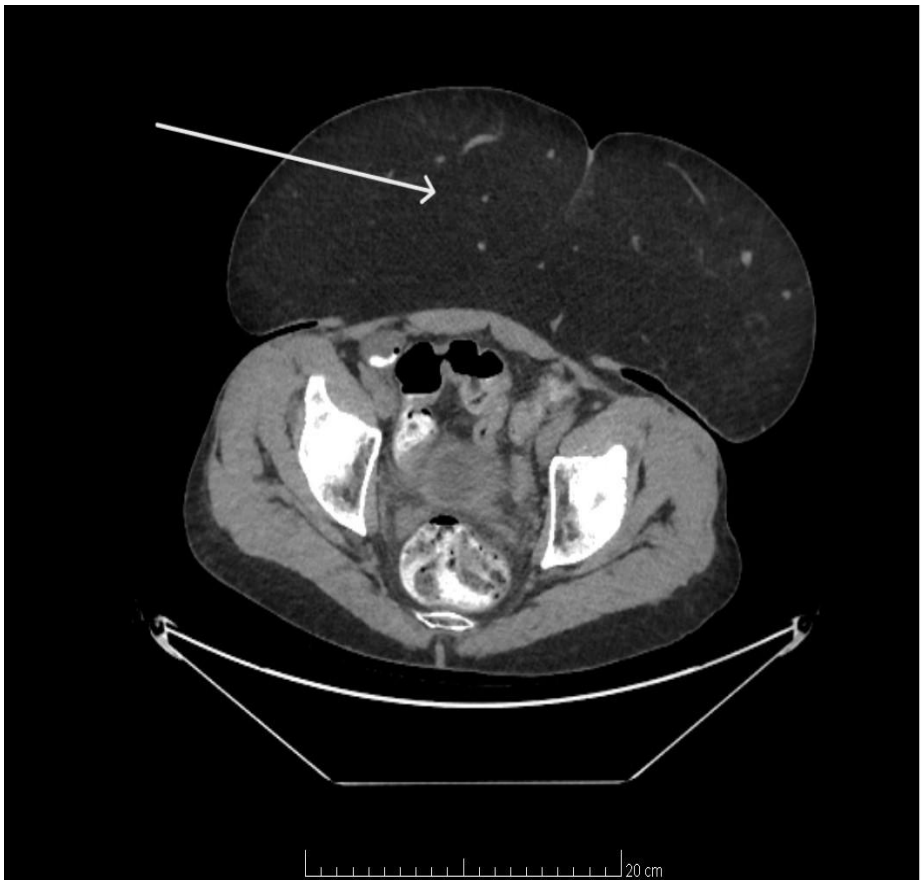

Fig. 2. Large amount of fat tissue in the computed tomography scan of the abdomen and minor pelvis

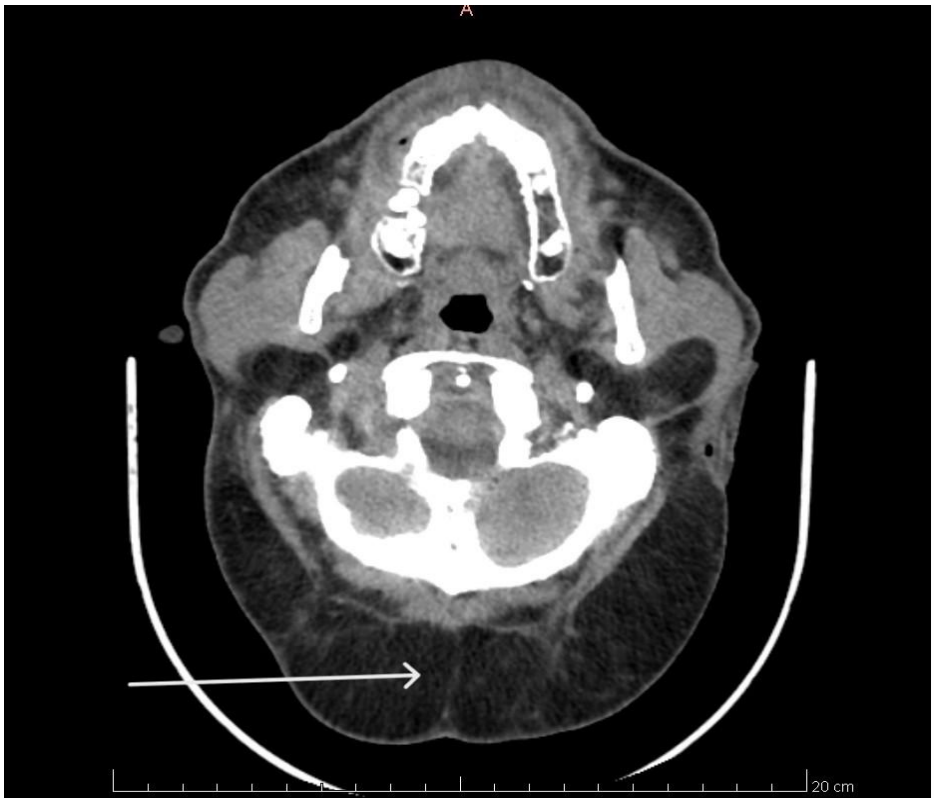

Fig. 3. Significantly thickened fatty tissue layer below the occipital bone on the computed tomography scan of the neck

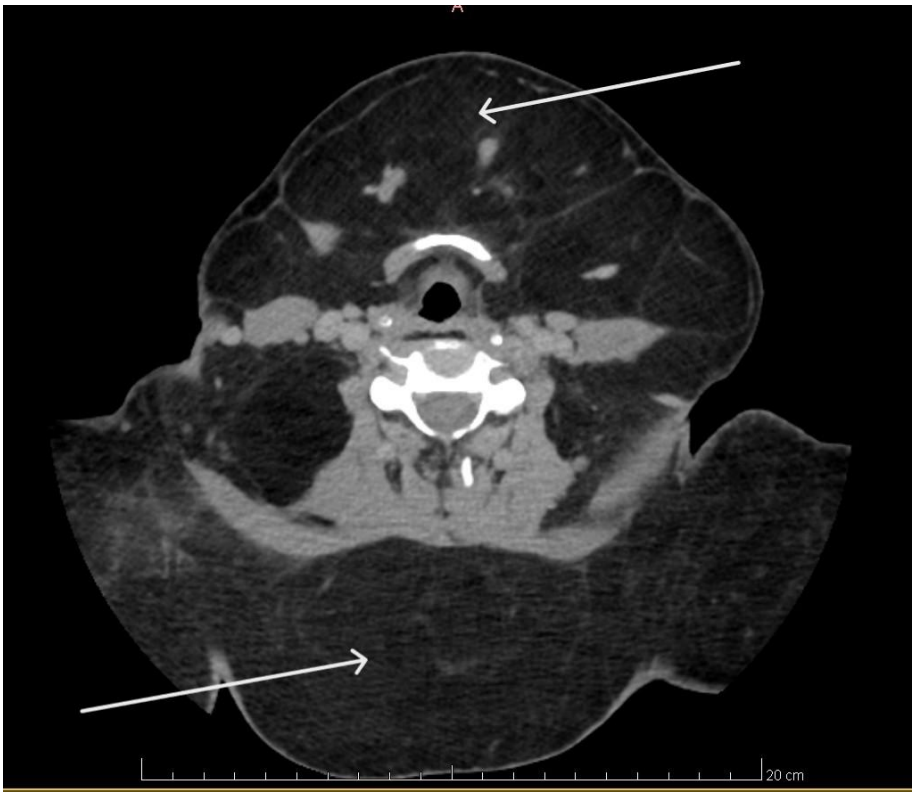

Fig. 4. Significantly thickened fatty tissue layer at the neck and forward from the hyoid bone on the computed tomography scan of the neck

Additional assessment of the cardiovascular system

Due to the reported dyspnea, elevated blood pressure during hospitalization, increased body mass, and atherosclerotic plaques on diagnostic imaging, an additional cardiovascular assessment was deemed necessary. Transthoracic echocardiography revealed slightly decreased left ventricular ejection fraction (51%). However, due to the suspicion of a bicuspid aortic valve, transesophageal echocardiography was conducted, confirming the presence of a rudimentary left coronary leaflet of the aortic valve. Carotid ultrasound revealed single atherosclerotic plaques bilaterally, none of which were hemodynamically significant. The intima-media thickness was 0.7 mm in the left carotid artery and 0.8 mm in the right carotid artery. On pulse wave analysis, the parameters of peripheral and central blood pressure were significantly elevated. Central pulse pressure was 66 mmHg. Pulse wave velocity on the femoral-carotid segment was within normal range (9.0 m/s). Ankle-brachial index and toe-brachial index on the left side were 0.99 and 0.71, respectively, and on the right side 0.98 and 0.64, respectively.

Further management

Based on the examinations and clinical symptoms, the patient was diagnosed with MD. After discharge from the hospital, the patient was advised to attend a scheduled consultation regarding plastic and reconstructive surgery. The patient was prescribed oral antibiotic therapy for *Helicobacter pylori* infection and the treatment for chronic hypertension was modified. Additionally, the patient was advised to obtain histopathological results of the samples collected during the endoscopic procedures.

DISCUSSION

MD is a rare condition, first described by Brodie in 1846 [4] and by Madelung in 1888 [5]. It is characterized by the progressive development of symmetrical, benign fatty tumors, which predominantly affect the upper body. These lipomas are commonly located around the neck, shoulders, upper arms, and trunk, giving patients a distinctive, pseudo-athletic appearance [1,2]. The disease is most frequently observed in middle-aged men, often with a history of chronic alcohol consumption [6,7]. Moreover, some reports describe an improvement in tumour mass after alcohol abstinence [8]. Additionally, patients with MD often present with metabolic abnormalities, including insulin resistance, dyslipidemia, and hypertension, which suggests a possible association with metabolic syndrome [1]. More unusual presentations, such as scrotal involvement, have been documented [9].

The pathophysiology of MD remains unclear; however, recent studies, such as that by Ma et al. [10], suggest that MD may be associated with altered adipocyte differentiation and an aberrant immune response, as indicated by single-cell RNA sequencing. This ana-

lysis provides deeper insights into the molecular mechanisms underlying the disease, particularly the role of immune cells in disease progression. The adipocytes in patients with MD are said to be brown adipose tissue cells [11]. However, the latest research suggests that genetic factors play a role in the pathogenesis of MD. A potential association between MD and mitochondrial DNA mutations, including mutation m.8344A>G and m.8357T>C in the *MT-TK* gene, has been identified in certain MD cases. The studies show that lipomas are rarely signs of mitochondrial disease, but in every MD case, mitochondrial DNA analysis should be considered [12,13,14,15]. Another mutation suggesting mitochondrial dysfunction in patients with MD is the mitofusin 2 (*MFN2*) mutation. *MFN2* is an essential gene for proper mitochondrial function and fusion, influencing many biological processes inside the cell, including lipid metabolism. Various pathogenic *MFN2* mutations have been identified in patients with MD through whole-exome sequencing [16,17] and Sanger sequencing [18]. Several other studies have proposed additional mutations that may underlie the development of MD symptoms, such as the Cbl proto-oncogene B (*CBLB*) mutation [19] or mutations in the gene encoding hormone-sensitive lipase (LIPE) [20].

The management of MD remains primarily surgical, with only additional and limited pharmacologic options (e.g., salbutamol or peroxisome proliferator-activated receptors-α agonists) [21]. However, surgical resection of lipomas does not yield long-term results in most cases and multiple removals are required. Lipectomy is often combined with liposuction of the fatty masses [22]. In turn, Scevola et al. [23] conducted a study of two patients with MD who underwent serial intralipotherapy with phosphatidylcholine/deoxycholate administered into the fatty lesions. On follow-up, they reported a 42.5% average reduction in lesion size across all treated lesions.

While MD is typically benign, rare complications can arise. One such complication is the transformation of benign lipomas into malignant forms, particularly liposarcomas. Lungu et al. [21] reported a case of MD progressing to liposarcoma, underscoring the importance of long-term surveillance for malignancy in these patients despite the risk of malignancy remaining low. Other rare complications include nerve compression, cervical immobility, or difficulties in breathing and dysphagia due to the size of the fatty masses [24,25]. Furthermore, there are reports of patients experiencing psychological distress due to the cosmetic impact of the condition, which can affect quality of life [26,27].

Despite MD's distinctive clinical signs, its rarity and physicians' relatively limited knowledge make it challenging to identify. In the case described herein, we have undertaken the entire diagnostic procedure necessary to establish the diagnosis, except for foreseeable complications. Unfortunately, there was no feedback from the patient after hospitalization and his subsequent course is unknown.

CONCLUSIONS

MD is a rare, complex disorder that presents both diagnostic and therapeutic challenges. Recent studies have provided valuable insights into its molecular and genetic basis, suggesting that its pathogenesis involves a combination of immune dysregulation, mitochondrial mutations, and environmental factors, such as alcohol consumption. Despite advances in understanding its molecular mechanisms, treatment remains primarily surgical, with limited pharmacologic options. As more research is conducted, it is hoped that new therapies will emerge and that a better understanding of the disease's pathophysiology will lead to improved diagnostic and treatment strategies, enhancing the quality of life for individuals with MD. Furthermore, as this disease may potentially progress to malignancy, continuous long-term follow-up is essential for affected individuals.

Authors' contribution

Study design – A. Basek, G.K. Jakubiak
Data collection – G.K. Jakubiak, A. Król-Zybura, M. Pietrzak, M. Starzak
Manuscript preparation – A. Basek, G.K. Jakubiak, G. Cieślar, A. Stanek
Literature research – A. Basek, G.K. Jakubiak
Final approval of the version to be published – A. Basek, G.K. Jakubiak, A. Król-Zybura, M. Pietrzak, M. Starzak, A. Stanek, G. Cieślar

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