Perineal Benign Symmetric Lipomatosis in a Female Patient: A Case Report

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Patient: Female, 33-year-old
Final Diagnosis: Benign symmetric lipomatosis
Symptoms: Mass in perineal region
Clinical Procedure: —
Specialty: Plastic Surgery

Objective: Unusual clinical course
Background: Benign symmetric lipomatosis (BSL), also known as Madelung’s disease, is a rare disease generally characterized by fat deposits painlessly and symmetrically distributed in the body. In former studies, the incidence of BSL is highest in male patients and more frequent in the Mediterranean area. Asian females seem to be rarely affected. BSL often occurs in the neck or upper-back of patients, and is related to various metabolic disorders. Unusual clinical appearances may add difficulties in diagnosis.

Case Report: In this report, we present a case of BSL in a 33-year-old woman’s perineal region, with no clear BSL risk factors (sex, medical history, sites, and comorbidities), which increased the difficulties in diagnosis. The patient’s quality of life was seriously affected by the continuous growth of fat tissue. Based on MRI and B-ultrasonic examinations, she underwent excision at our outpatient facility. Combined with the patient’s clinical appearance, imaging results, and pathological tests, we could finally determine the diagnosis of BSL. After 18 months of follow-up, this patient recovered well with no recurrence.

Conclusions: Difficulties in diagnosis can seriously affect doctors’ treatment approaches. BSL rarely occurs in the lower body, and our patient showed no clear risk factors. Therefore, imaging and pathological examinations can be essential tools for dermatological and plastic surgeons to diagnose and treat rare BSL.

Keywords: Case Reports • Diagnosis • Madelung Deformity

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Introduction

Benign symmetric lipomatosis (BSL) – also known as Madelung’s disease, multiple symmetric lipomatosis, or Launois-Bensaude syndrome – is a rare lipid metabolic disorder first reported by Brodie [1]. According to previous studies, BSL is more common in men than in women, generally affects middle-aged men in the neck or upper-back area, and is related to alcohol abuse [2,3]. However, to date, few cases have been reported in women and in non-drinkers. In this case report, we present a rare case of BSL in a young woman with an uncommon tumor region and no clear risk factors.

Case Report

A 33-year-old woman (height: 173 cm; weight: 65 kg; BMI: 21.72 kg/m²) with a giant mass in her perineal area visited our outpatient clinic in June 2022. The perineal mass, first occurring 7 years ago, was initially located in the patient’s pubic region. Thereafter, the mass started growing gradually, with no symptoms of discomfort, from the pubic to the rear region on both sides of the body. The bilateral masses rubbing against each other due to continuous growth seriously affected her quality of life. As a result, she sought treatment.

In November 2021, the patient underwent radical resection for left thyroid cancer (postoperative pathological reports revealed papillary thyroid cancer), and she recovered well. Since then, she has been regularly taking levothyroxine sodium tablets (100 μg qd) and maintaining stable normal thyroid function. She reported regular menstruation and stated she had no other previous illnesses, or any history of smoking or alcohol abuse.

Symmetric giant masses were found in the patient’s perineal region, and they were approximately 20×8 cm in size with no skin ulceration and exudation (Figure 1). Due to mutual friction, the surface skin color of the mass in the contact area was slightly reddened. The mass was soft, with no tenderness.

Figure 1. Clinical appearance of the patient (A-C) and the excised tissue (D).
Examinations of the patient’s routine blood and urine test results, as well as liver function, kidney function, blood lipid levels, and coagulation function, indicated no significant abnormalities. Besides thyroid-stimulating hormone (TSH: 0.044 μIU/ml, reference values: 0.380-4.340 μIU/ml), she showed overall normal thyroid function.

We first based our diagnosis of BSL on the symptoms the patient manifested and reports of physical examinations; however, owing to her gender, negative medical history, and the uncommon tumor site, we completed more imaging examinations, including MRI and B-ultrasound, to reconfirm our diagnosis. Imaging examinations not only helped us determine the nature of the BSL mass, but also provided more information on mass growth and guidance on treatment. B-ultrasound found perineal subcutaneous fat-like echoes with unclear borders (Figure 2). MRI results revealed symmetrically distributed thickening and swelling subcutaneous soft tissue in the perineum, with high-signal areas both in T1-mapping and in T2-mapping (fat-suppression images showed low-signal areas), while the structures of surrounding muscles and bones were normal (Figure 3).

Based on the above-described clinical evidence, we confirmed our diagnosis of BSL and performed treatment combining mass excision and liposuction. The surgery was successful, and the mass was removed (Figure 1) and sent for pathological examination. The excised mass was lobulated without a complete capsule. Pathological images showed proliferated fat tissue (Figure 4). After 18 months of follow-up, this patient had recovered well, with no signs of recurrence.

Discussion

A review study [4] reported that BSL is a rare disease, which mostly occurs in males (male-to-female ratio: 15-30: 1), and nearly 50% of BSL patients are aged 30-60 years. All ethnic groups can be affected, but BSL is more common in Mediterranean areas and rare in Asia [2]. Female cases are also rarely reported, but they can be affected [2,5]. The mechanisms of BSL are still unclear. According to a study by Li [6], 81.48% of patients had endocrinal diseases, including hyperlipidemia (33.33%), hyperuricemia (29.63%), diabetes (25.93%),
Figure 3. MRI images of the mass. (A) T1-mapping of the mass. (B) Soft-tissue mass of high-signal intensity. (C) T2-mapping of the mass. Soft-tissue mass of high-signal intensity with low-signal intensity fibrous septa. T2-mapping of fat suppression. Soft-tissue mass showing low-signal intensity.
and obesity (16.67%). In this respect, metabolic disorders may play a significant role in BSL development. Moreover, 89.5% of patients reported alcoholism [4], which may be considered as an important risk factor. Few studies have analyzed the causes of the low incidence of BSL among females. In China, males may be more susceptible to alcoholism and metabolic disorders than females [7]. Therefore, we inferred that lifestyles and habits may be responsible for the low incidence rate of BSL among females. In our case report, the BSL-positive female patient showed no clear risk factors and had undergone radical resection for left thyroid cancer in 2021. It was important for us to consider the relationship between the patient’s thyroid function history and BSL, but she reported no history of abnormal thyroid function.

BSL is generally found in the neck or upper-back region; however, cases of BSL on the proximal ends of the limbs have also been reported. In 2002, Nounla [8] reported a perineal BSL case, which has since remained rarely reported.

In the female BSL case discussed in this paper, the signs the patient showed and the reports of physical examinations confirmed BSL; however, more imaging examinations were necessary for a complete diagnosis and treatment recommendation due to the uncommon gender and tumor site. Notably, there were no noticeable risk factors or any related comorbidities. Zhang [9] summarized the results of MRI imaging of BSL patients. In T2-mapping, fat showed high-signal intensity with low-signal fibrous septa; however, the fat showed low-signal intensity in T2-mapping with fat suppression. CT is also an effective examination for evaluating the nidus. However, we did not choose CT as a regular examination due to its radioactivity. Additionally, B-ultrasound is an effective approach to evaluating superficial masses, especially their blood supply situation. Prior research did not describe in detail the results of B-ultrasound for BSL. In this case report, B-ultrasound reported subcutaneous fat-like echoes with unclear borders and a low blood flow signal. Although pathological examinations are considered the criterion standard for diagnosis of tumors other than BSL, such examinations are still necessary and reveal proliferated fat tissue. Generally, BSL is diagnosed via physical and histological examination, combined with imaging analysis [10].

A common approach to BSL treatment is lipectomy, liposuction, or a combination of these. Dermatological or plastic surgeons, based on imaging results, can recommend the best treatment modality for the patient. In this study, the patient received combined therapy. After 1 year of follow-up, the patient has recovered well, with no recurrence.

Conclusions

In dermatology and plastic surgery, some rare diseases may have a typical clinical appearance; moreover, patients may report unusual medical history, sites, or comorbidities. Relevant imaging examinations may offer comprehensive information on the evaluation of masses, which can provide diagnostic value and guide dermatological surgeons and plastic surgeons with regard to treatment.

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