

Benign Symmetric Lipomatosis (Madelung's Disease): A case report of new variant

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ABSTRACT

Benign symmetric lipomatosis (BSL) is characterized by the presentation of numerous symmetric non-tender fatty tumors in the neck, suboccipital region, proximal extremities and upper parts of the trunk, typically noted as a "pseudoathletic" appearance. The etiology of BSL remains unknown but mitochondrial mutations have been found in some patients. The mitochondrial cytopathy was suggested to be studied in all BSL patients especially those with neuropathy. We have presented a case of BSL with reverse fat distribution, which we named the "Pop-eye" appearance in which the fatty tumor was distributed at both the distal part of the upper extremities and the proximal part of the lower extremities. A study of the mitochondrial tRNA lysine gene did not reveal mutation at position 8344.

Keywords: Benign symmetric lipomatosis, Madelung's disease, "Pop-eye" appearance

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CASE REPORT

Benign symmetric lipomatosis (BSL or Madelung's disease) is a rare disorder characterized by the presentation of numerous symmetric non-tender fatty tumors in the neck, suboccipital region, proximal extremities and upper parts of the trunk. BSL was first described by Brodie in 1846 and named after Madelung in 1888.¹ Subsequently, multiple synonyms for this order have been used, such as multiple symmetrical lipomatosis, benign lipomatosis, lipomatosis simplex, Ekbohm's syndrome and Madelung's disease. BSL is usually found in adults, age 30-60 years, with an incidence of 1:25,000 and M:F ratio of 15:1 to 30:1.¹ Mitochondrial mutations have been found in some patients.² More than 90% of patients suffer from alcoholism and the symptoms may be associated with some metabolic abnormalities.² Our case was diagnosed as Madelung's disease with a reverse distribution of the lipomas and no mutation of mitochondrial tRNA lysine gene.

A 34-year-old Thai man presented with progressively enlarged arms and legs for 4 years. The growth of extremities troubled both walking and social functions. He had been diagnosed with epilepsy since he

was 8 years old and his symptoms had to be controlled by Phenytoin (300 mg/day) and Phenobarbital (120 mg/day). He had no history of alcohol abuse, and none of his family members were affected by this condition.

The physical examination showed a symmetrical enlargement of the distal part of the upper extremities and the proximal part of the lower extremities, including his buttocks. The neck and trunk were of normal shape (Fig 1). There were a large café-au-lait patch at the right posterior thigh and old surgical scars from drainage of an abscess when he was young at his left posterior thigh and left buttock. The motor power of all extremities was in grade V with normal deep tendon reflexes. He also had strabismus in his left eye. Other systems are within the normal limits. His complete blood count, fasting blood sugar, electrolytes, renal function, liver function and thyroid function, lipid profiles and urine examinations were normal. A magnetic resonance imaging (MRI) study revealed non-encapsulated fat tissue in both thighs (Fig 2).

A muscle biopsy specimen taken from the left biceps revealed non-specific change and no evidence of ragged-red fibers. Molecular genetic analysis studied shows no mutation of the mitochondrial lysine transfer RNA gene at position 8344.

The patient was treated with liposuction and the fat tissue was histologically revealed as mature fat cells without inflammation.

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Fig 1. A symmetrical enlargement of the distal part of the upper extremities and the proximal part of the lower extremities, including his buttocks showing "Pop-eye appearance".

DISCUSSION

Benign symmetric lipomatosis was characterized by Madelung in 1888 and Launos and Bensaude in 1898³; as symmetric non-tender fatty tumors in the neck, suboccipital region, proximal part of extremities and the upper part of the trunk. Enzi delineated two types of BSL: a type I pseudoathletic appearance and a type II generalized obese appearance.⁴ Our patient had symmetric non-tender, non-encapsulated, fatty masses which were localized at distal parts of the upper extremities and proximal parts of lower extremities giving the "Pop-eye" appearance. Our case was found to have no metabolic abnormalities which have been previously reported.^{2,5} A café-au-lait patch has never been reported with BSL but it can be found in 10-20% of the normal population,⁶ therefore, the only café-au-lait patch in our patient could be a co-incidental finding.

The pathophysiology of BSL is unknown, but may involve an alteration in mitochondrial gene expression affecting brown fat cell proliferation and differentiation.^{2,5} Muscle biopsy is the gold standard to diagnose mitochondrial disease by checking for the 8344mt DNA mutation which can be found in 17% of BSL patients (Myoclonus Epilepsy Associated with Ragged-Red fibers syndrome; MERRF syndrome).⁷ Our case neither showed CNS involvement nor mtDNA mutations as evidence of the MERRF syndrome. The diagnosis of BSL can be established with clinical presentation; imaging studies such as ultrasonography, computer tomography or

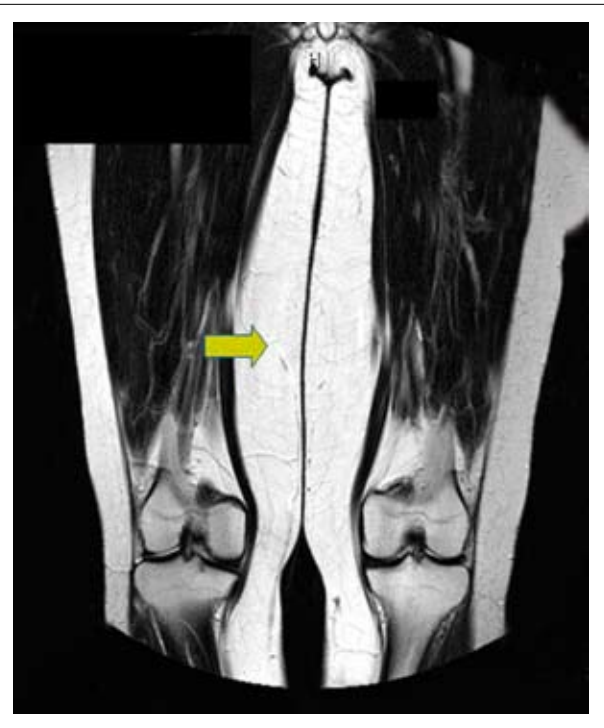


Fig 2. Coronal-sectional MR T2-weighted imaging showed high signal intensity (arrow) which represent fatty tissue. This figure showed excess symmetric unencapsulated subcutaneous at both thighs.

magnetic resonance imaging may be useful^{8,9} as in this case.

The treatment for BSL is surgical removal of the fatty tissue (open approach and/or liposuction) to improve the quality of life of the patients.^{1,9,10} However, the fatty tissues usually reappear in a matter of time. There are reports of using various medical treatments such as intralesional injection of enoxaparin, fenofibrates or salbutamol, as an alternative therapy for BSL but they have shown inconsistent results.^{6,11-13}

In conclusion, we present a BSL case with new, unusual fatty tumor distributions, molecular genetic analysis for the mitochondrial lysine tRNA gene at position 8,344, and imaging, which was treated by surgical removal of the fatty tumor.

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