中文題目：罕見疾病 Madelung's disease 的臨床表現：一病例報告與文獻回顧
英文題目：Madelung's disease — A Case Report
作　　者：羅澤賢[1]、張士彥[1]、簡紹南[2]、徐北辰[1]
服務單位：苗栗大千綜合醫院[1]內科部、[2]影像醫學部

Introduction:

Madelung's disease (MD), also known as Launois–Bensaude syndrome, is a rare disorder of lipid metabolism. The fat deposition is symmetrically distributed mainly around the shoulders, neck and upper trunk, but some patients may appear at hips and thighs. Most of the patients are asymptomatic condition except painless swelling formation. The disease is first documented by Brodie in 1846. The German surgeon Otto Madelung reported the first series of 33 patients with fat neck in 1888.

Case report:

A 53-year-old man presented to our medicine out-patient department because of left arm and bilateral thigh swelling for more than five years. He did not feel pain, tense or itchy. He is a patient of chronic hepatitis C, had received interferon therapy and completed treatment three years ago. He has chronic alcohol drinking habit for about 30 years. Daily drinking amount is two bottles of rice wine. He did not play body-building exercise anytime. He felt gradually increased of left arm size, so that he visited to our out-patient department. Physical examination demonstrated swelling and masses of left arm, bilateral shoulders, lower back and bilateral thighs. These swelling were soft and not tendered when we palpated. The laboratory blood tests showed mild liver enzyme elevation but white blood cell counts, CRP, ANA, rheumatoid factor, cholesterol and LDL levels were all within normal range. Magnetic resonance imaging (MRI) of left arm revealed high signal intensity on T1 weighted image, low signal intensity on short T1 inversion recovery (STIR) or fat suppressant weighted image. This MRI finding was compatible with lipomatosis of left arm. No biopsy was done.

Discussion:

The symmetric enlargement of shoulders, arms and back (Pseudo-bodybuilding appearance) without regular weight-lifting is very rare and it may relate with lipomatosis. Patients with MD are middle-aged (30~60 years old) and mostly found in Mediterranean countries. Men is more affected than women. Vassallo GA et al described three types of MD. Type 1 is distributed mainly around the shoulders, neck and arms. Type 2 mainly occurs at upper back, deltoid area, hips and thighs. Type 3 is associated with gynecoid distribution of lipomatosis. The etiology is still unknown but

it may be due to dysfunction of lipid metabolism. Catecholamine-stimulated lipolysis impairment may play an important role. Perera U et al stated that the Canadian patients with MD carry MTTK gene c.8344A>G variant in mitochondrial DNA. Most patients are associated with chronic alcohol drinking. Hyperlipidemia, hyperglycemia, hypothyroidism and liver disease may also be present. Our patient has drinking habit and chronic hepatitis C with interferon treatment. But the clinical correlation between MD and anti-viral treatment has not been established. Most of the patients present with painless soft tissue swelling and very few clinical manifestations, and mainly face with cosmetic problem. The disease may be under-diagnosed due to lack of symptoms. Computed tomography or MRI reveal lipomatosis. The important differential diagnoses include liposarcoma, multiple familial lipomatosis, neurofibroma, Cushing syndrome and angiolipoma. Aspiration cytology may help confirmation, but is rarely needed. The diagnosis of MD is based on clinical and imaging examinations. Although there is no pharmacological treatment, alcohol withdrawal and weight loss are recommended. The treatment options in severe cases are liposuction and lipectomy. Liposuction becomes more popular due to less invasive and better cosmetic outcomes. Our patient did not have any discomfort except cosmetic problem. We suggested abstinence from alcohol and conservative management. No surgical intervention was arranged.