REVIEW

1761 Cardiac rehabilitation and its essential role in the secondary prevention of cardiovascular diseases
Winnige P, Vysoky R, Dosbaba F, Batalik L

ORIGINAL ARTICLE

Case Control Study

1785 Association between homeobox protein transcript antisense intergenic ribonucleic acid genetic polymorphisms and cholangiocarcinoma

Retrospective Study

1793 Risk factors for post-hepatectomy liver failure in 80 patients
Xing Y, Liu ZR, Yu W, Zhang HY, Song MM

1803 Outcomes of laparoscopic bile duct exploration for choledocholithiasis with small common bile duct
Huang XX, Wu JY, Bai YN, Yu JY, Lv JH, Chen WZ, Huang LM, Huang BF, Yan ML

Observational Study

1814 Three-dimensional finite element analysis with different internal fixation methods through the anterior approach
Xie XJ, Cao SL, Tong K, Zhong ZY, Wang G

1827 Bedside cardiopulmonary ultrasonography evaluates lung water content in very low-weight preterm neonates with patent ductus arteriosus
Yu LF, Xu CK, Zhao M, Niu L, Huang XM, Zhang ZQ

CASE REPORT

1835 Conservative endodontic management using a calcium silicate bioceramic sealer for delayed root fracture: A case report and review of the literature
Zheng P, Shen ZY, Fu BP

1844 Brain magnetic resonance imaging findings and radiologic review of maple syrup urine disease: Report of three cases
Li Y, Liu X, Duan CF, Song XF, Zhuang XH

1853 A three-year clinical investigation of a Chinese child with craniometaphyseal dysplasia caused by a mutated ANKH gene
Wu JL, Li XL, Chen SM, Lan XP, Chen JJ, Li XY, Wang W

1863 Intradural osteomas: Report of two cases
Li L, Ying GY, Tang YJ, Wu H
Thrice Monthly Volume 9 Number 8 March 16, 2021

1871  Gastroesophageal varices in a patient presenting with essential thrombocythemia: A case report  

1877  Chest pain showing precordial ST-segment elevation in a 96-year-old woman with right coronary artery occlusion: A case report  
   Wu HY, Cheng G, Cao YW

1885  Subcutaneous panniculitis-like T-cell lymphoma invading central nervous system in long-term clinical remission with lenalidomide: A case report  
   Sun J, Ma XS, Qu LM, Song XS

1893  Imaging findings of primary pulmonary synovial sarcoma with secondary distant metastases: A case report  
   Li R, Teng X, Han WH, Li Y, Liu QW

1901  Severe community-acquired pneumonia caused by *Leptospira interrogans*: A case report and review of literature  
   Bao QH, Yu L, Ding JJ, Chen YJ, Wang JW, Pang JM, Jin Q

1909  Bilateral common peroneal neuropathy due to rapid and marked weight loss after biliary surgery: A case report  
   Oh MW, Gu MS, Kong HH

1916  Retroperitoneal laparoscopic partial resection of the renal pelvis for urothelial carcinoma: A case report  

1923  17α-hydroxylase/17,20 carbon chain lyase deficiency caused by p.Tyr329fs homozygous mutation: Three case reports  

1931  Epithelioid angiomyolipoma of the pancreas: A case report and review of the literature  

1940  Computed tomography imaging features for amyloid dacryolith in the nasolacrimal excretory system: A case report  
   Che ZG, Ni T, Wang ZC, Wang DW

1946  Epidural analgesia followed by epidural hydroxyethyl starch prevented post-dural puncture headache: Twenty case reports and a review of the literature  
   Song LL, Zhou Y, Geng ZY


1968  Human parvovirus B19-associated early postoperative acquired pure red cell aplasia in simultaneous pancreas-kidney transplantation: A case report  
<table>
<thead>
<tr>
<th>Year</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>1976</td>
<td>Diabetes insipidus with impaired vision caused by germinoma and perioptic meningeal seeding: A case report</td>
<td>Yang N, Zhu HJ, Yao Y, He LY, Li YX, You H, Zhang HB</td>
</tr>
<tr>
<td>1983</td>
<td>Madelung disease: A case report</td>
<td>Chen KK, Ni LS, Yu WH</td>
</tr>
<tr>
<td>2001</td>
<td>Inadvertent globe penetration during retrobulbar anesthesia: A case report</td>
<td>Dai Y, Sun T, Gong JF</td>
</tr>
<tr>
<td>2015</td>
<td>Interstitial lung disease induced by the roots of Achyranthes japonica Nakai: Three case reports</td>
<td>Moon DS, Yoon SH, Lee SI, Park SG, Na YS</td>
</tr>
</tbody>
</table>
ABOUT COVER
Gokul Sridharan, MD, PhD, Associate Professor, Oral Pathology and Microbiology, YMT Dental College and Hospital, Navi Mumbai, Mumbai 400018, Maharashtra, India. drgokuls@gmail.com

AIMS AND SCOPE
The primary aim of World Journal of Clinical Cases (WJCC, World J Clin Cases) is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

INDEXING/ABSTRACTING
The WJCC is now indexed in Science Citation Index Expanded (also known as SciSearch®), Journal Citation Reports/Science Edition, Scopus, PubMed, and PubMed Central. The 2020 Edition of Journal Citation Reports® cites the 2019 impact factor (IF) for WJCC as 1.013; IF without journal self cites: 0.991; Ranking: 120 among 165 journals in medicine, general and internal; and Quartile category: Q3. The WJCC’s CiteScore for 2019 is 0.3 and Scopus CiteScore rank 2019: General Medicine is 394/529.

RESPONSIBLE EDITORS FOR THIS ISSUE
Production Editor: Jia-Hui Li; Production Department Director: Yu-Jie Ma; Editorial Office Director: Jin-Lei Wang.

NAME OF JOURNAL
World Journal of Clinical Cases

ISSN
ISSN 2307-8960 (online)

LAUNCH DATE
April 16, 2013

FREQUENCY
Thrice Monthly

EDITORS-IN-CHIEF
Dennis A Bloomfield, Sandro Vento, Bao-Gan Peng

EDITORIAL BOARD MEMBERS
https://www.wjgnet.com/2307-8960/editorialboard.htm

PUBLICATION DATE
March 16, 2021

COPYRIGHT
© 2021 Baishideng Publishing Group Inc

INSTRUCTIONS TO AUTHORS
https://www.wjgnet.com/bpg/gerinfo/204

GUIDELINES FOR ETHICS DOCUMENTS
https://www.wjgnet.com/bpg/gerinfo/287

GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH
https://www.wjgnet.com/bpg/gerinfo/240

PUBLICATION ETHICS
https://www.wjgnet.com/bpg/gerinfo/288

PUBLICATION MISCONDUCT
https://www.wjgnet.com/bpg/gerinfo/208

ARTICLE PROCESSING CHARGE
https://www.wjgnet.com/bpg/gerinfo/242

STEPS FOR SUBMITTING MANUSCRIPTS
https://www.wjgnet.com/bpg/gerinfo/239

ONLINE SUBMISSION
https://www.f6publishing.com
Madelung disease: A case report

Ke-Ke Chen, Lian-Song Ni, Wei-Hui Yu

ORCID number: Ke-Ke Chen 0000-0002-9846-4489; Lian-Song Ni 0000-0002-1097-2474; Wei-Hui Yu 0000-0001-7384-0113.

Author contributions: Chen KK collected clinical data, discussed the details, contributed to the writing; Ni LS contributed to the follow-up of the patient, and modified the manuscript; Yu WH modified the manuscript and was a major contributor in writing the manuscript.

Informed consent statement: Informed written consent was obtained from the patient for publication of this report and any accompanying images.

Conflict-of-interest statement: The authors declare that they have no conflict of interest.

CARE Checklist (2016) statement: The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

Open-Access: This article is an open-access article that was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution NonCommercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build

Key Words: Madelung disease; Multiple lipomatosis; Diabetes; Case report

©The Author(s) 2021. Published by Baishideng Publishing Group Inc. All rights reserved.

Core Tip: Madelung disease (MD) is a rare disorder of fat metabolism, which results in diffuse and symmetrical deposition of adipose tissue in the subcutaneous superficial fascia space and/or deep fascia space in various parts of the body, such as the neck and shoulders, etc. Recently, a case of MD accompanied by type 2 diabetes was diagnosed and treated in our hospital. The clinical characteristics of MD are still under-recognized. Herein we discuss the pathogenesis, diagnosis and treatment of MD in order to improve the clinician’s understanding of the disease.

Abstract

BACKGROUND
Madelung disease (MD) is a rare disorder of fat metabolism, resulting in diffuse, symmetrical and painless deposition of adipose tissue in subcutaneous superficial fascial space and/or deep fascia space of the head, neck and shoulders, etc.

CASE SUMMARY
We report a case of MD accompanied by type 2 diabetes in a 61-year-old Chinese male. The patient presented with progressive fat deposition over the mandible, neck, abdomen and elbows. He had a history of smoking and alcohol abuse. Excessive fat deposition was seen in the mandible, elbows and the abdominal area of the patient by ultrasonic examination. Computed tomography showed diffuse and marked soft masses (fat density) in the subcutaneous superficial fascia space of the neck. The patient was diagnosed with MD. He was advised to abstain from alcohol and was followed up regularly.

CONCLUSION
This report discusses the pathogenesis, diagnosis and treatment of MD, and raises the clinician’s awareness of this disease.

Ke-Ke Chen, Lian-Song Ni, Wei-Hui Yu, Department of Endocrinology, The First Affiliated Hospital of Wenzhou Medical University, Wenzhou 325000, Zhejiang Province, China

Corresponding author: Wei-Hui Yu, MD, PhD, Attending Doctor, Department of Endocrinology, The First Affiliated Hospital of Wenzhou Medical University, No. 2 Fuxue Alley, Wenzhou 325000, Zhejiang Province, China. shadow1201@163.com
INTRODUCTION

Madelung disease (MD) is a relatively rare metabolic syndrome with unclear etiology. The main characteristic of the disease is the diffuse, symmetrical, and painless deposition of adipose tissue in the subcutaneous superficial fascial space and/or deep fascia space of the neck, shoulders and proximal extremities, etc[1-3]. It usually occurs in middle-aged men with a history of chronic alcoholism. The disease is more common in the Mediterranean population, and very rare in the Asian population[2]. This report describes a recent case of MD accompanied by type 2 diabetes who was diagnosed and treated in our hospital.

CASE PRESENTATION

Chief complaints
A 61-year-old male of Han ancestry was admitted to our hospital on July 15, 2020 due to a 4-yr history of progressive swollen soft masses at multiple sites.

History of present illness
The patient presented with progressive soft masses over the abdomen for more than four years, swellings in the mandible, back of the neck and elbows also gradually enlarged during these four years.

History of past illness
The patient was a heavy drinker with an average of 500 mL liquor per day for 45 years. He had a smoking history of approximately 20 cigarettes/day for 45 years,. The patient was diagnosed with type 2 diabetes more than 20 years previously, with poorly controlled glucose for many years. He had a history of hypertension for 18 years and did not take any antihypertensive drugs.

Personal and family history
He had no family history of similar diseases and denied previous special drug use.

Physical examination
Physical examination upon admission revealed the following: The patient was conscious; a body mass index of 21.0 kg/m²; subcutaneous masses in the mandible, back of the neck, abdomen and elbows (Figure 1); no signs of moon face, paper skin, hirsutism, or purple stria.

Laboratory examinations
Laboratory tests showed a total cholesterol level of 92.16 mg/dL, low-density lipoprotein cholesterol level of 35.28 mg/dL, high-density lipoprotein cholesterol level of 37.44 mg/dL, triglyceride level of 20.7 mg/dL, hemoglobin A1c of 5.4% and 24 h urinary albumin was 343.5 mg/d. Thyroid function was in the normal range. His basal cortisol level as well as his 1 mg dexamethasone suppression test were normal. Hepatitis B, hepatitis C, human immunodeficiency virus and syphilis were all negative.

Imaging examinations
Ultrasonography showed diffuse lipomatosis and lymph node echo in the neck in January 2018. The thickest part of the mass in the neck was about 28 mm. In July 2020, ultrasonography showed that the thickest part of the mass in the neck was about 44 mm. Ultrasonography of the elbow suggested that there was a hypoechoic mass between the palmaris muscles on both sides of the elbow, with a range of about 51 mm × 15 mm × 34 mm on the left and 46 mm × 14 mm × 35 mm on the right. The boundary was clear and the internal echo was not uniform, with strong echoes of several fiber
spacers, similar to the echoes of adipose tissue. Simultaneously, excessive fat deposition was suggested on the mandible and the abdominal area of the patient by ultrasonic examination. Computed tomography showed diffuse and marked fatty deposits in the subcutaneous superficial fascia space of the neck without internal lesions (Figure 2). A breast ultrasound examination showed that both sides of the breast had excessive fat deposition and a flaky strong echo was seen in the deep side of the areola. The left side was about 33 mm × 3 mm × 25 mm and the right side was about 44 mm × 8 mm × 31 mm. The internal lobular structure was disordered, and small hypoechoic areas were seen, but no obvious mass echo and mammary duct expansion was found.

**FINAL DIAGNOSIS**

The patient was diagnosed with MD according to history taking, clinical features and auxiliary examinations.

**TREATMENT**

The patient was prescribed glimepiride, metformin, and voglibose to control blood glucose, and losartan potassium to reduce blood pressure and albuminuria. Alcohol abstinence and follow-up were advised. Surgical treatment was not considered at this time.

**OUTCOME AND FOLLOW-UP**

The patient was in a stable condition at follow-up visit. Alcohol abstinence and follow-up every 3-mo were advised.

**DISCUSSION**

MD is a disorder of fat metabolism. Patients with MD mainly show diffuse, symmetrical, painless and irreversible deposition of adipose tissue in the subcutaneous superficial fascia space and/or deep fascia space in the neck, shoulder, back and proximal limbs, and is a benign disease [1-4]. The disease is also referred to as benign or multiple symmetrical lipomatosis (MSL), or the Launois-Bensaude syndrome. Previous reports have shown that the disease is more frequent in Mediterranean men, and more common in middle-aged people, aged 30-60 years old, with a male to female ratio of 15:1 to 30:1 [5]. The incidence of MD is rare, and it is not difficult to diagnose according to the patient’s history and physical signs. However, if clinicians do not know enough about MD, it is easily missed or misdiagnosed. Based on the anatomical location of adipose tissue, MD can be categorized into two types. Type I is the most common type, and found mainly in males. The adipose tissue is mainly distributed in the upper body, such as upper trunk, cervical region, supraclavicular region and arms. The area of fat in the neck slowly expands, giving it the appearance of a “horse collar”.

![Figure 1 Fat deposition in different parts of the body in a 61-year-old man with Madelung disease. A: Mandible and back of neck; B: Elbow; C: Abdomen.](https://www.wjgnet.com)
Patients are admitted to hospital mainly due to “painless neck mass and progressive enlargement”. These patients usually present with concomitant symptoms including weight loss. In contrast, Type II MD, which is often accompanied by weight gain and unrelated to alcoholism, can also occur in females. The fat deposits mainly in the upper back, deltoid muscle area, buttocks and upper thigh, and some patients have upper abdominal fat accumulation\(^6\) and individual cases of pediatric patients have previously been reported\(^7,8\). The diagnosis of MD is predominantly based on history taking, physical examination and auxiliary examinations. Ultrasound shows symmetric subcutaneous fat layer thickening, involvement of part of the muscle layer, an unclear boundary, irregular shape, often visible cord-like echoes, and the blood supply is not rich. The characteristic findings on CT or MRI play significant roles in the diagnosis and differential diagnosis\(^9\). Although the pathogenesis is still unclear, MD is considered an autosomal dominant inherited disease\(^10\). Nisi et al\(^11\) considered that MD is caused by a local defect of lipolysis caused by mitochondrial dysfunction. MD is strongly associated with alcohol consumption, which directly affects mitochondrial activity, causing premature oxidation of mitochondrial DNA or a point mutation in the tRNA-lysine gene of mitochondrial DNA (A8344G)\(^12\). Musumeci et al\(^13\) confirmed that MSL could be considered, even if rare, a red flag for mitochondrial disorders, even in patients with an apparently isolated MSL. It has also been suggested that it may be related to brown fat hypertrophy caused by functional sympathetic degeneration of adipose tissue, which is the result of an abnormality in the synthesis of intracellular cyclic adenosine monophosphate induced by the stimulation of noradrenaline, the lipogenic, antilipolytic, and decreased lipid oxidative effect of ethanol may play a permissive role in the growth of fat in susceptible individuals\(^14\). On the other hand, findings have shown that MFN2-related MSL is a novel mitochondrial lipodystrophic syndrome involving both lipomatous masses and lipoatrophy\(^15\). The development of MD may also be associated with abnormal glucose metabolism, hyperlipidemia, hypothyroidism, peripheral neuropathy of lower limbs, myoclonic epilepsy with ragged-red fibers syndrome and hyperuricemia\(^16,17\). To date, there is no effective drug therapy for MD. It has been reported that, in a few cases, the fat mass in patients will subside with abstinence and weight loss\(^18\). Therefore, the general treatment of MD is reduced to palliative treatment, including abstinence from alcohol and diet control. For most patients, discontinuation of alcohol abuse and a bariatric procedure may help control the disease, but has little effect on inhibiting progression of the disease. Surgical resection of the adipose tissue, liposuction, or injection lipolysis is still the most effective treatment, although patients often relapse after treatment\(^19\). Brea-García et al\(^20\) reported a total postoperative recurrence rate of 63%, nevertheless, up to 95% of patients experienced symptoms relapse after liposuction\(^21\). Therefore, regular long-term clinical follow-up of patients is recommended.
CONCLUSION

MD, a rare disease that progresses relatively slowly and is known as benign or multiple symmetrical lipomatosis, often coexists with a variety of metabolic disorders. This case introduces the involvement of alcohol abuse in MD pathogenesis. There is currently no effective treatment for the disease; therefore, it is important that patients are regularly followed up. At present, the clinical characteristics of MD are still under-recognized. We hope that this article can contribute to increasing the understanding of MD.

ACKNOWLEDGEMENTS

We are grateful to the patient, who gave his informed consent for publication.

REFERENCES


