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Case Report

Massive labial lipomatous hypertrophy in familial partial lipodystrophy seen on computed tomographic angiography

Kevin Selting, BS, Neil J. Hansen, MD, W.L. Harrison, MD*

University of Nebraska College of Medicine, Department of Radiology, 981045 Nebraska Medical Center, Omaha, NE 68198-1045

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ABSTRACT

We present a 28-year-old female with a rare familial partial lipodystrophy. Originally presenting at the age of 14, she began experiencing hypertrophy of the fat in the mons pubis and labia majora regions. By the age of 24 she had disfiguring hypertrophy of these areas with severe fatty overgrowth, similar in nature to that experienced by her father and paternal grandmother. During her workup and planning for suction lipectomy, she underwent computed tomography angiography with the imaging manifestation of severe massive subcutaneous fat hypertrophy; the imaging appearance was only able to be explained after a thorough review of the patient's history and medical literature.

Case report

A 14-year-old Caucasian female with a noncontributory past medical history presented with cervical lipomatosis. Similar dysmorphic features were noted in her father and paternal grandmother, leading to a genetics evaluation. By the time she was 24, she carried the diagnosis of Familial Partial Lipodystrophy (FPLD) type 2. On physical examination, she was noted to have a prominent mandible with submental and cervical lipodystrophy both anteriorly and posteriorly. She had marked prominence of her mons pubis and labia majora with severe subcutaneous fat hypertrophy. She had confirmatory molecular genetic testing/gene sequencing (autosomal dominant mutation in the LMNA gene).

Given her symptoms from the labial lipohypertrophy, she presented to a Plastic Surgeon for surgical correction. A computed tomography angiogram (CTA) of the abdomen and pelvis was performed to evaluate the vasculature of the pelvis and perineal region. Her abnormal fatty tissue proliferation was not readily evident on prior radiographs or the scout image (Fig. 1). Massive lipomatous hypertrophy of the labia majora and mons pubis was noted (Figs. 2 and 3). No mass, enhancement, or infiltration of the fat were identified, nor were any other lipomatous lesions. No lymphadenopathy or vascular abnormalities were noted.

Declaration of Competing Interest: None of the authors have financial or other conflicts of interest.

* Corresponding Author.
E-mail address: njhansen@unmc.edu (N.J. Hansen).

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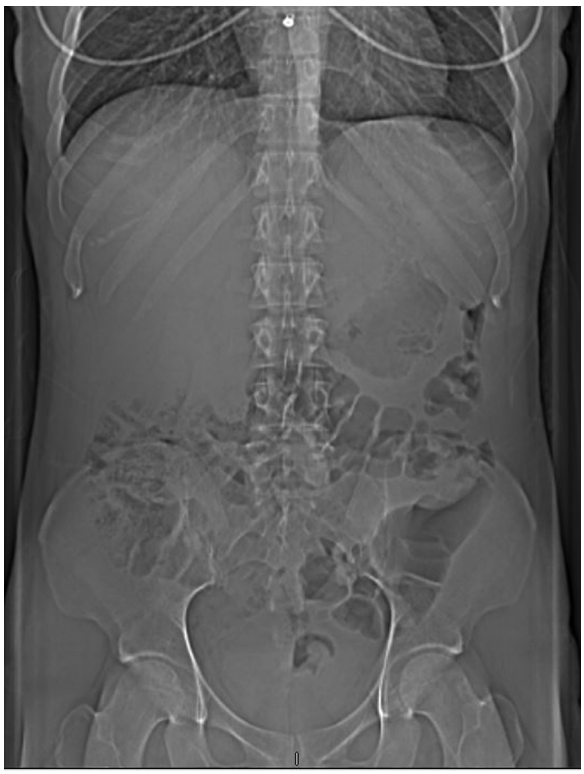

Fig. 1 – Scout image from computed tomography angiography (CTA) of the abdomen and pelvis. The massive lipomatous hypertrophy over the mons pubis region is not readily discernable.

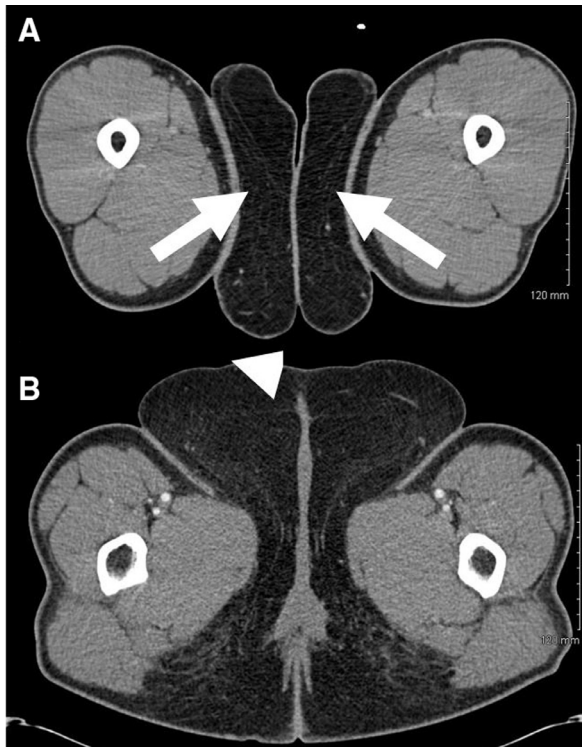

Fig. 2 – Axial contrast enhanced computed tomography angiogram (CTA) images of the pelvis revealed massive lipomatous hypertrophy of the labia majora (Arrows, A) and mons pubis (Arrowhead, B). No mass, enhancement, or infiltration of the fat were identified, nor were any other lipomatous lesions. No lymphadenopathy or vascular abnormalities were noted.

The patient then underwent a 2-stage operation involving a suction lipectomy followed by excision of excess skin. Her postoperative course was complicated by recurrent seroma, which eventually resolved.

Discussion

FPLD syndromes are rare disorders of variable lipoatrophy in certain body regions and normal or excess adiposity in others [1–5]. By far, the most common lipodystrophy is antiretroviral-induced lipodystrophy in HIV patients [2,6]. Aside from iatrogenic causes, the differential for lipodystrophy includes congenital generalized lipodystrophy, FPLD, acquired generalized lipodystrophy, and acquired partial lipodystrophy [2]. These conditions are extremely rare with the former 2 reported in approximately 500 patients worldwide and the latter 2 found in less than 30. There is almost nothing in the literature regarding imaging of patients affected by these disorders. It may be the radiologist, when encountering such unusual imaging manifestations as those seen in Fig. 1, who is the first provider to consider such a rare diagnosis. Radiologists must be aware of the imaging manifestations found in patients known to have these disorders, so as not to mistake lipohypertrophy for a fat containing tumor.

Through clinical findings and genetic testing, our patient was found to have FPLD type 2, which is due to an

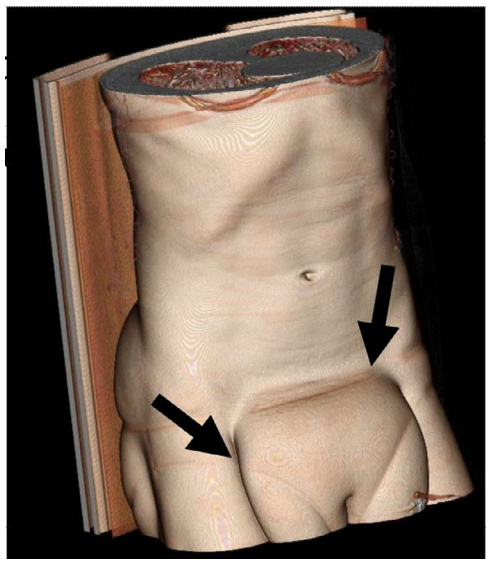

Fig. 3 – Three-dimensional Volume rendered image demonstrating disfigurement related to massive hypertrophy of the fat in the mons pubis/labia majora regions (arrows).

autosomal dominant heterozygous missense mutation in the LMNA gene, responsible for encoding nuclear lamin proteins A and C. A hyperinflammatory state caused by endoplasmic reticulum stress is also thought to play a role in the pathogenesis of type 2 FPLD, or Dunnigan type lipodystrophy [7]. Patients characteristically will develop diabetes and dyslipidemias, which predisposes them to early cardiovascular disease [8].

The initial treatment for FPLD is aimed at treating the metabolic derangements associated with the condition. The treatments are the same for those without dyslipidemias. Persistent metabolic disturbances can be treated with metreleptin, an analog of human leptin that has showed modest efficacy [9,10].

Imaging in the context of lipodystrophies is yet to be completely characterized. It has been noted that radiotracer will accumulate in subcutaneous and visceral fat following Fluorine-18 Fluorodeoxyglucose Positron Emission Tomography/Computed Tomography in patients with HIV-induced lipodystrophy. This could have important effects on the pharmacodynamics of certain drugs, altering medical management [11]. The use of echocardiography in this patient population can help to identify epicardial adipose tissue [8]. As we saw in our patient with Dunnigan type FPLD, computed tomography angiogram was used to further characterize the vascularity of the lipomatous hypertrophy for preoperative planning. Anatomic evaluation of the vasculature involved was important to the 2-stage operation that followed. This also allowed the surgeon to rule out lymphatic, infective, vascular, and other etiologies that may have been accounting for the mons and labial enlargement given the extreme rarity of this disorder and unique clinical presentation. At the time of surgical planning, the exact composition of the physical abnormality in the mons region was in need of better anatomic delineation.

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