Ultrasonographic Manifestations of Multiple Tuberous Xanthomas on Bilateral Buttocks with Hypercholesterolemia: One rare case report

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Key Clinical Message

Xanthomas is a rare benign disorder. This lesion is often associated with endocrine and metabolic diseases (e.g. diabetes mellitus, hyperlipidemia, etc) and characterized by deposition of lipid-laden histiocytes. We report one rare case of multiple tuberous xanthomas on bilateral buttocks with hypercholesterolemia. The patient visited doctors for palpable masses on his bilateral buttocks without any pain. In this case, ultrasonic(the iU22 scanner (Philips Healthcare, Andover, MA) equipped with a 12-5 MHz linear-array transducer.) manifestation revealed hyperechoic masses located in the skin and subcutaneous tissues of bilateral buttocks, and blood flow signals were abundant within the masses, and low-velocity, low-obstruction artery and low-velocity venous blood flow spectrum were detected. The patient was subsequently operated for surgical removal. The masses were all excised under local anaesthesia. The diagnosis was finally confirmed by postoperative pathologic examination. In conclusion,ultrasonic characteristic manifestation of multiple tuberous xanthomas on bilateral buttocks with hypercholesterolemia can provide important information for the diagnosis of this rare case.

Key words: multiple tuberous xanthomas, hypercholesterolemia, bilateral buttocks, ultrasonography

IntroductionXanthoma is a rare yellow-to-skin-colored benign lesion in patients with endocrine and metabolic diseases. [1] Multiple tubercular xanthomas of buttocks is also a rare complication of familial hypercholesterolemia. [2-4] And multiple tubercular xanthomas on bilateral buttocks with hypercholesterolemia are even an extremely rare lesion. Xanthomas are palpable masses that are typically located within the skin or subcutaneous tissue and consist of cholesterol, triglycerides, phospholipids, and numerous lipid-laden foamy macrophages. [5] It has been reported in previous literatures that xanthomatosis mainly occurs in eyelid, elbow, tendon, bone, and other parts, and it is more common in young children and middle-aged women. [6,7]We reported an extremely rare case of bilateral buttocks xanthomas in a 31-year-old man. His last follow-up 5 years after surgery showed no sign of recurrence in clinical and ultrasonography (US) examination. Generally, the value of ultrasound diagnosis lies in defining the scope, layer, and depth of the lesion. Thereby, ultrasonic findings have certain auxiliary value for selection of surgical resection method.

2. Case presentation

The patient, a 31-year-old male, was found several hemispheric nodules in the skin or subcutaneous of both buttocks, disease course of 5 years, and these nodules were slightly soft. The patient did not feel painful and itching in his buttocks, and the skin of buttocks was intact. The masses of bilateral buttocks had significantly increased in the past 1 month. The case underwent mass resection and pathological examination, and recovered well with no relapse within 5 years after surgery.

Physical examination : By palpation, we found all masses with poor mobility, moderate texture and no fluctuation.

Laboratory examination : Through laboratory tests, we found that serum triglyceride was 1.62 mmol/L (normal value 0.29-1.83mmol/L), low-density lipoprotein was 11.2 mmol/L (normal value $<$ 4.0mmol/L), high-density lipoprotein was 1.51 mmol/L (normal value $>$ 0.9 mmol/L), and cholesterol was 16.7 mmol/L (normal value 2.8-5.7 mmol/L).

Ultrasonic findings : We found that multiple hyper echoic masses were in the skin layer of bilateral buttocks, and the biggest mass of left buttock was about 69×14×44 millimeter in size, with clear boundary and irregular morphology (Fig. 1). Abundant blood flow signals were seen within the masses, and low-velocity, low-obstruction artery and low-velocity venous blood flow spectrum were detected (Fig. 2).

Postoperative pathological specimens : Two masses were about 31×15×30 millimeter and 30×25×30 millimeter in size on right buttock respectively, and a lump was about 69×14×44 millimeter in size on left buttock. The sizes of above masses were all consistent with that measured by ultrasound.

Pathological findings : Histopathologic lesions of masses on left and right buttocks were analyzed by immunohistochemical and histochemical staining. The masses were mainly composed of a large number of focal-like foam cells, which were nodular in distribution(Fig. 3A). Inflammatory cell infiltration, fibroblast hyperplasia with a small amount of cholesterol deposition and hyaline degeneration were seen in some areas(Fig. 3A). Immunohistochemic: S-100 (-), Leu-7 (-), Desmin (-), SMA (-), inhibin (-), CD68 (+), which supported the pathological diagnosis with xanthomas (Fig. 3B).

3.Discussion

Xanthomatosis of the skin is a group of diseases characterized by yellow patches, papules, or nodules, accompanied by increased lipids and other organ abnormalities. [8] Due to the accumulation of lipid-containing cells in the skin dermis, yellow nodular lesions often form on the skin surface. [9] The rash forms and distribution of this disease vary, and there are mainly 4 types: nodular, flat, rash and tendinous.[10] This disease can be associated with abnormal lipid metabolism. Some cases have familial or systemic lesions. For example, lipid deposition in the cardiovascular system that can produce arteriosclerosis and embolism of small vessels. [11,12]

There were few literature reports about ultrasonic findings of multiple tubercular xanthomas on bilateral buttocks with hypercholesterolemia. [13,14]And most of them is all about ultrasonic findings of achilles tendon xanthoma.[15,16]However, this case report is about ultrasonic characteristic manifestations of multiple tuber-

cular xanthomas on bilateral buttocks with hypercholesterolemia. Ultrasound can find the size, depth, layer of skin involved and blood supply of lesion, providing vital help for surgical options.

More importantly, Sonographic appearance had certain characteristics in this case. There were several hyperechoic masses in the skin dermis of bilateral buttocks, and the biggest was on the left buttock, with clear boundary and irregular shape. Abundant blood flow signals could be detected within the masses. Low-velocity, low-obstruction arterial and low-velocity venous blood flow spectra were detected, which might be a characteristic performance in diagnosing this rare case. Appearance of blood signal may indicate foam cell activity. Further study with a large sample size is needed to confirm it. Moreover, the uneven internal echo was meshed, which might be related to the nodular distribution of foam cells. Therefore, the ultrasonic characteristic findings contained uneven meshed internal echo and abundant blood flow signals within the masses, with low-obstruction arterial and low-velocity venous blood flow spectra. They can help to differentiate xanthomas from other diseases such as skin keloid in which blood flow signals cannot be detected. Due to the rarity of xanthomas at this site, the experience of ultrasonic examination still needs to be accumulated in future case study.

Author Contributions

Dongmei Li : Writing – review and editing; investigation

Lei Wang : Data curation; Writing – review and editing; Funding acquisition;

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Figure legends

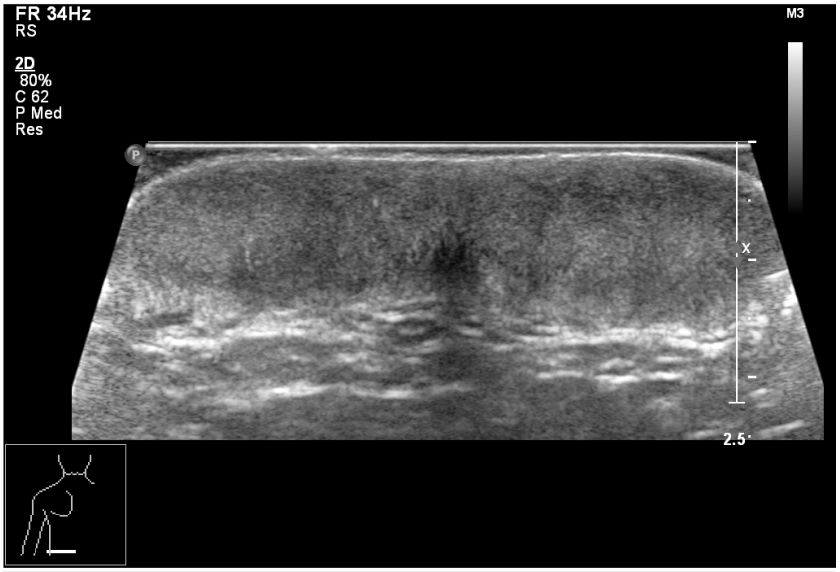
Fig.

1: Hyperechoic mass in the skin layer of the left buttock (the biggest mass) by the transversal axis (Fig. 1A) and longitudinal axis(Fig. 1B) of ultrasound

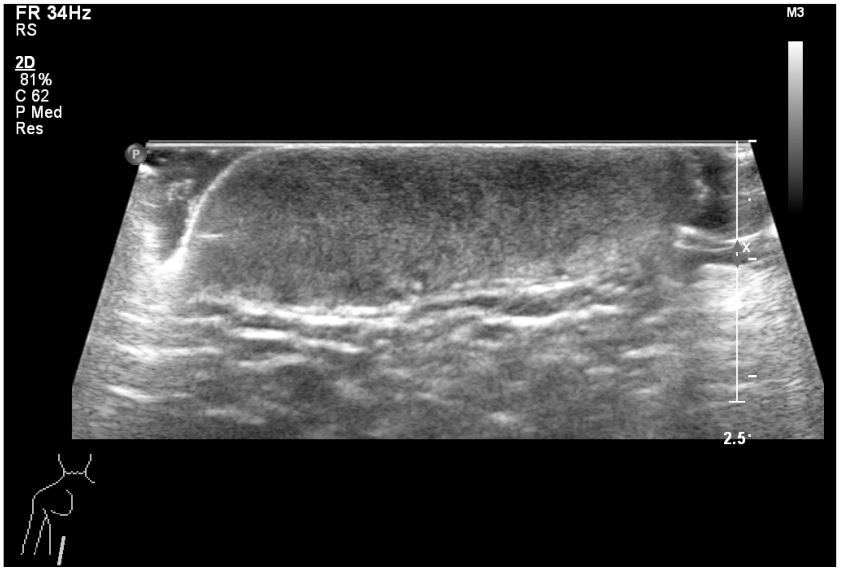
Fig. 2: Abundant blood flow signals seen within the mass (Fig. 2A), and low-velocity, low-obstruction artery blood flow spectrum detected from blood signals(Fig. 2B).

Fig. 3: Haematoxylin-eosin staining revealing an abundant foamy macrophages with fibrosis and cholesterol clefts(original magnification×400) (Fig. 3A), and Immunohistochemistry staining revealing a positive expression pattern: S-100 (-), Leu-7 (-), Desmin (-), SMA (-), inhibin (-), CD68 (+) (Fig . 3B)

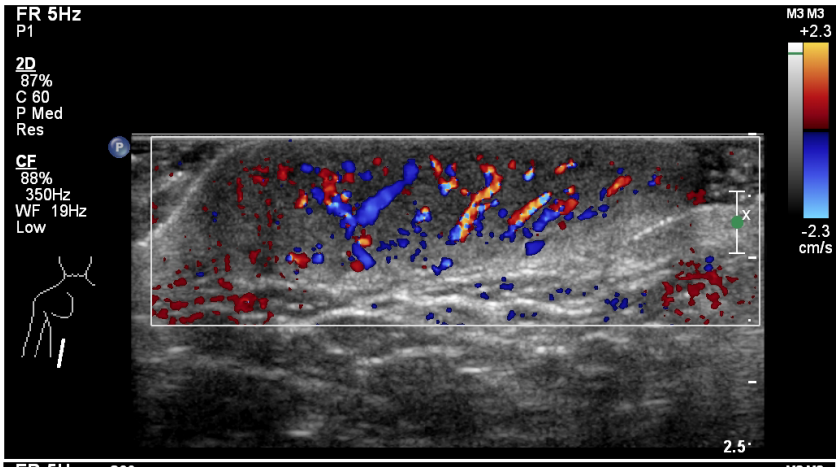
A



B



A



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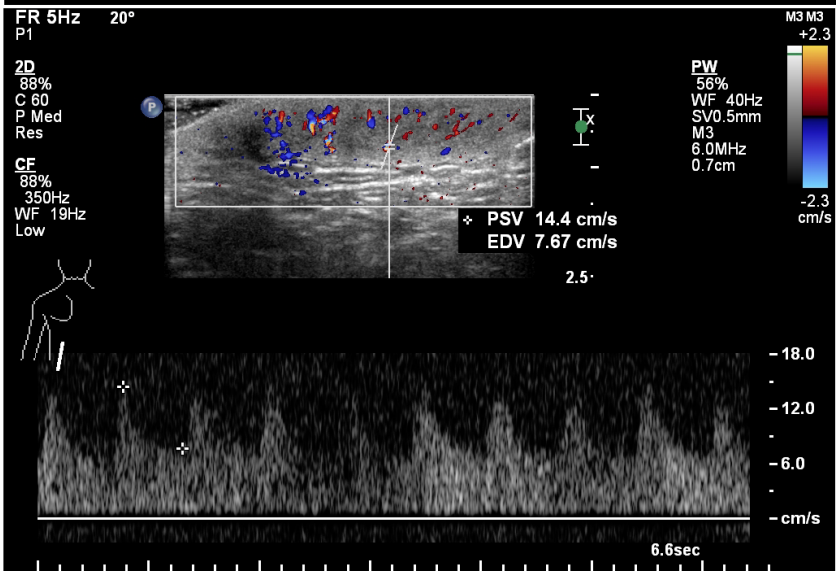

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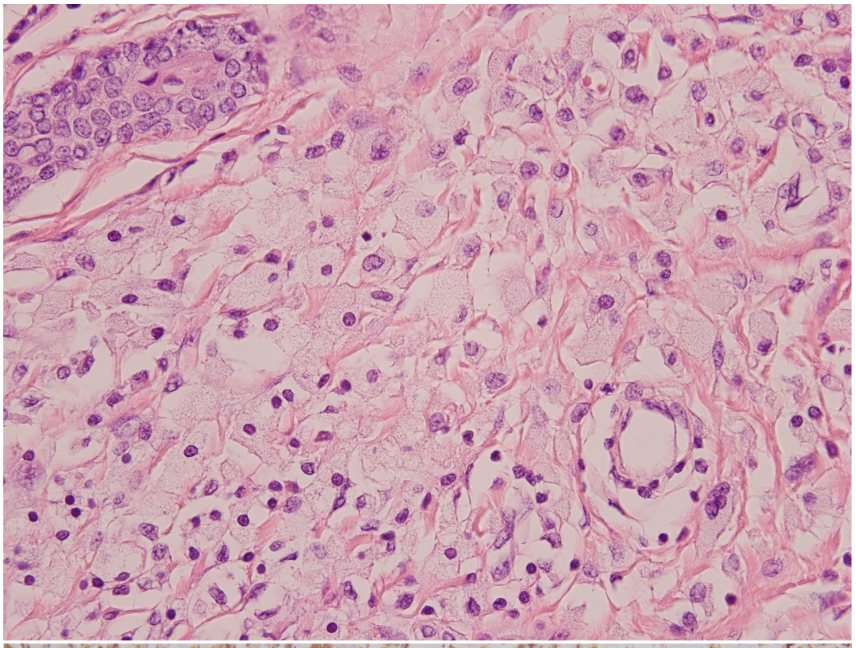

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