Diabetes mellitus and congenital proximal femur hypoplasia: is there a link or association

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We present a case of congenital short femur with scleredema Diabeticorum (SD) in a patient with diabetic neuropathy with unusual skin lesion distribution. Gait symptoms progressively worsened. The atypical distribution of the skin lesions is quite unusual. This case express rare complications of diabetes mellitus, the maternal diabetes which had disrupted in part the development of skeletal system of the offspring who in turn developed a rare dermatologic complications i.e. scleredema Diabeticorum (SD).
This case will leave inedible memory for every diabetologist not to miss the skeletal and dermatologic manifestations of DM. The combination of this congenital proximal femoral hypoplasia with complicating diabetes make the question if there is a link with similar genetic predilection or just an association.

Keywords:
atypical, congenital short femur, link, scleredema diabeticorum

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Introduction

Scleredema diabeticorum (SD) can be considered as a major dermatological complication of diabetes mellitus (DM). Histologically, there is a remarkable deposition of mucin along the deep dermis layers [1,2].

The typical skin lesions are ill-defined, nonpitting, erythematous, indurated plaques, with 'peau d'orange' appearance and its distribution is mainly in the upper part of the body – in the neck, trunk, and upper limbs. Face is frequently involved. The histologic findings of scleredema include deposition of mucin between dermal collagen bundles. The deposition is greatest in the deep dermis [3].

The etiopathogenesis is not clear, although the increased expression of collagen-producing fibroblasts in the skin of affected individuals may be preceded by or associated with a history of an antecedent febrile illness, DM, or blood dyscrasia. Other internal organ involvement has been reported [4].

Congenital short femur syndrome is a developmental disorder of the proximal segment of the femur leading to shortening of limb with abnormal gait. The etiology is unknown, but different theories are postulated, may be due to local vascular damage to mesenchymal tissue or intrauterine compression of the thigh at the time of femoral diaphysis ossification [5].

Case report

A 25-year-old woman with a diabetic mother and a history of type 1 DM for 12 years, noncompliant to treatment and married but infertile presented to us. Her parents noticed shortening of her lower limbs only with waddling gait since childhood; rapidly progressive thickening of skin was observed 3 months ago.

She presented to us with uncontrolled DM; she was mentally fair, her weight was 80 kg, height was 127 cm, BMI was 49.6, and span measured 143 cm, whereas the upper segment was 78.5 cm and the lower segment was 48.5 cm indicating affection of only LL (Fig. 1).

On examination, she had central obesity with a waist circumference of 116 cm. The patient had stock pattern hypothesia with generalized diffuse thickening and induration of the skin affecting the neck, trunk, upper extremities, and both thighs, which is a rare involvement. She had developed secondary sexual characters. Fundus examination showed bilateral moderate nonproliferative diabetic retinopathy.

Her laboratory investigations were normal apart from hyperglycemia, dyslipidemia, and microalbuminuria. Thyroid-stimulating hormone was normal, whereas

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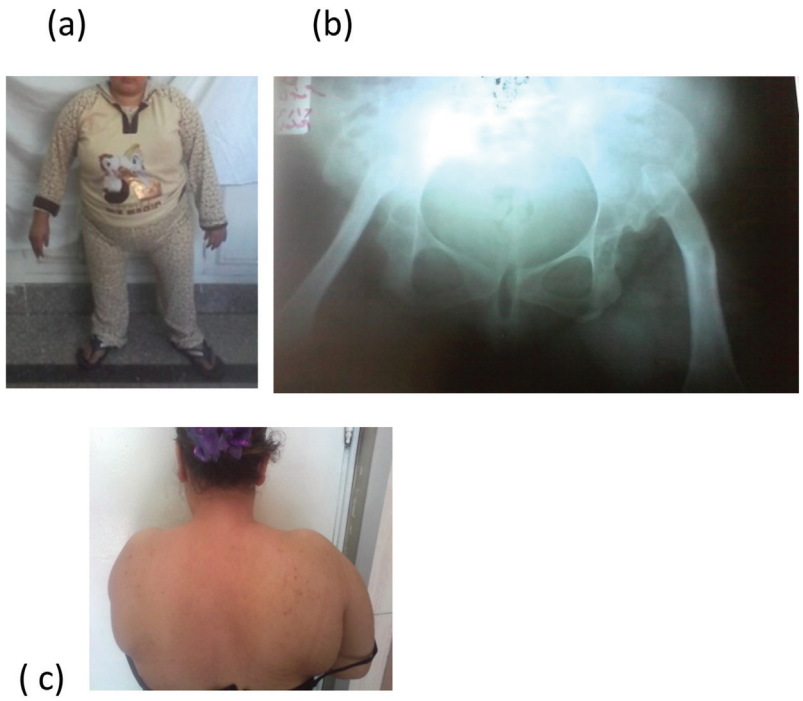
Figure 1



(a–c) The patient is short and obese with lateral bowing of both femora. Radiological imaging demonstrated bilateral hip dislocation with proximal femoral head and neck deficiency with shallow acetabulum. Sclerodema diabeticurum affecting the neck and the back.

follicle-stimulating hormone and luteinizing hormone were high for correlating phase indicating premature ovarian failure. Serum protein electrophoresis showed an increase in α-2 globulin region. Abdominal and pelvic sonoraphy revealed marked abdominal distension with multiple ovarian cysts indicating polycystic ovaries.

Skeletal radiography survey demonstrated bilateral hip dislocation with proximal femoral head and neck deficiency with shallow acetabulum (type III). Echocardiography and computed tomography of the abdomen were normal. This study has been approved from the ethical committee in the faculty.

Discussion

Proximal femoral deficiency or congenital short femur is a rare congenital bony anomaly manifested by failure of normal development of a variable portion of proximal femur with functional abnormality. A radiological classification proposed by Aitken [6] is used in diagnosis and management.

SD is a dermatologic complication of long-term DM that is usually considered to be benign; most of the times, it is not modified with metabolic control, thereby ensuing relentless deterioration.

Hitti *et al.* [7] were the first to present a detailed histopathological examination of aborted fetus with congenital femoral hypoplasia of a diabetic mother. The primitive mesenchyme cells derived from the lateral plate are destined to form the skeleton and tendons of the lower limb through transformation to a cartilaginous model that can undergo endochondral ossification to form the tubular shape of the femur. Hitti *et al.* [7] showed that at certain segment or focus of the diaphysis of the growing femur during embryogenesis the mesenchyme cells skip the cartilaginous transformation and get transformed into bone directly through intermembraneous ossification; this bony part is more or less flat, lacking the normal tubular architecture of the rest of femur, and hence the name focal femoral deficiency. It is known that the cartilaginous development is sensitive to the level of glucose [8]. The abnormal glucose level selectively inhibit cartilage-specific proteoglycan core protein gene.Interestingly, mesenchyme cell differentiation into cartilage depends on extracellular matrix and cell adhesion molecules. Similarly, the pathogenesis of SD is attributed mainly to aberrant expression of extracellular matrix genes in skin fibroblast [2].

Conclusion

Our case is scleredema diabiticorum presenting with congenital short femur with progressive symmetric indurations of the skin affecting unusual sites (both thighs) with no history suggestive of internal organ

affection such as dysphagia, breathing or cardiac complication or arrhythmias.

This is an uncommon case of progressive SD with congenital proximal femoral dysplasia.

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Conflicts of interest
There are no conflicts of interest.

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