

DM & congenital proximal femur hypoplasia: is there a link or association

Abstract

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 in two faces, first 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. 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 makes the question if there is a link with similar genetic predilection or just an association.

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

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, diabetes mellitus, 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 aetiology is unknown, but different theories are postulated, may be due to local vascular damage to mesenchymal tissue or intrauterine compression of the thigh at time of femoral diaphysis ossification.⁵

Case report

A 25-year old female of diabetic mother with history of being diabetic for 12years, non-compliant to treatment, married but she is infertile. Her parents noticed shortening of her lower limbs only with waddling gait since childhood, she observed rapidly progressive thickening of skin 3months ago. She was presenting to us with uncontrolled DM, she is mentally fair, her weight is kg, height is 127cm, and BMI is span measures 143cm while upper segment 78.5cm & lower segment 48.5cm indicating affection of only LL (Figure 1).

On examination, the patient had stock pattern hypothesis with generalized diffuse thickening & induration of the skin affecting the neck, trunk, upper extremities& both thighs which is a rare

Volume 6 Issue 4 - 2017

Mervat Essam, Ahmed Yamani

Lecturer of internal medicine, Faculty of medicine, Cairo University, Egypt

Correspondence: Mervat Essam, Lecturer of internal medicine, Faculty of medicine, Cairo University, Egypt, Email lambaa2020@yahoo.com

Received: August 25, 2016 | **Published:** April 06, 2017

involvement (Figure 2). Fundus examination showed bilateral diabetic changes.



Figure 1 A 25-year old female of diabetic mother.

Her labs were normal apart from hyperglycemia, dyslipidemia & microalbuminuria. TSH was normal while FSH & LH were high for correlating phase indicating pre mature ovarian failure. Protein electrophoresis showed increase in alpha 2 globulin region. Abdominal & pelvic sonography revealed marked abdominal distension with multiple ovarian cysts indicating polycystic ovaries (PCO). Skeletal x-ray survey demonstrated bilateral hip dislocation with proximal femoral head & neck deficiency with shallow acetabulum (type III) (Figure 3). Echocardiography & CT abdomen were normal.



Figure 2 Fundus examination showed bilateral diabetic changes.



Figure 3 Echocardiography & CT abdomen were normal.

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 is employed in diagnosis and management.⁶ 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.⁷ 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, 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.

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 internal organ affection such as dysphagia, breathing or cardiac complication or arrhythmias. This is an uncommon case of progressive scleredema diabiticorum with congenital proximal femoral hypoplasia.

Acknowledgements

None.

Conflict of interest

The author declares no conflict of interest.

References

1. Farrell AM, Branfoot AC, Moss J, et al. Scleredema diabiticorum of Buschke confined to the thighs. *Br J Dermatol.* 1996;134(6):1113–1115.
2. Varga J, Gotta S, Li L, et al. Scleredema adulorum: Case report and demonstration of abnormal expression of extracellular matrix genes in skin fibroblasts *in vivo* and *in vitro*. *Br J Dermatol.* 1995;132(6):992–999.
3. Kokpol C, Rajatanavin N, Rattanakemakorn P. Successful treatment of scleredema diabiticorum by combining local puva and colchicine: a case report. *Case Rep Dermatol.* 2012;4(3):265–268.
4. Tran K, Boyd KP, Robinson MR, et al. Scleredema diabiticorum. *Dermatol Online J.* 2013;19(12):20718.
5. Levinson ED, Ozonoff MB, Royen PM. Proximal femoral focal deficiency (PFFD). *Radiology.* 1977;125:197–203.
6. Aitken G. Proximal femoral focal deficiency. Definitions, classification and management. In: Aitken GT editor. *Proximal focal femoral deficiency, a congenital anomaly.* A symposium, USA: National Academy of Sciences; 1968.
7. Hitti, Ibrahim F. Bilateral femoral hypoplasia and maternal diabetes mellitus: case report and review of the literature. *Pediatric Pathology.* 1994;14(4):567–574.†
8. Leonard CM, Bergman M, Frenz DA, et al. Abnormal ambient glucose levels inhibit proteoglycan core protein gene expression and reduce proteoglycan accumulation during chondrogenesis: Possible mechanism for teratogenic effects of maternal diabetes. *Proc Natl Acad Sci USA.* 1989;86(24):10113–10117.