

DOI: 10.21767/2471-8041.100048

The First Case of Cutis Laxa Type II (Debre Type) Associated with Atrial Septal Defect

Aamir J Al-Mosawi

Children Teaching Hospital, Medical City, Israel

Corresponding author: Aamir J Al Mosa wi, Senior Advisor and Doctor , Children Teaching Hospital, Medical City, Israel, Tel: 00964770390834; E-mail: almosawiAJ@yahoo.com**Rec Date:** March 10, 2017, **Acc Date:** April 16, 2017, **Pub Date:** April 18, 2017**Citation:** Al-Mosawi AJ. The First Case of Cutis Laxa Type II (Debre Type) Associated with Atrial Septal Defect. Med Case Rep, 3:2.

Abstract

Cutis laxa type II (MIM 219200) is a rare congenital disorder, first described by Debre et al. in 1937. During the period from 1937 to 2008, there have been 34 (25 females and 9 males) well-documented published cases of this rare disorder.

In 2009, I reported the thirty fifth case of this disorder and I also reviewed the previously published and thirty four well documented cases. A dysmorphic girl was referred at the age of Four years (early April 2016) because of skin abnormality and delayed speech. The girl had cutis laxa with redundant loose skin present from birth. This skin abnormality was evident on the limbs and abdomen. Dysmorphic facial feature included, broad and depressed nasal bridge, and the eyes were wide set with down-slanting palpebral fissures. Examination of the cardiovascular system was normal except for the presence of a systolic murmur. Echocardiography revealed the presence of a small atrial septal defect.

In this paper, I am reporting the 36th case of Cutis laxa (Debre type) which is the first case to be associated with atrial septal defect.

Keywords: Cutis laxa (Debre type); Atrial septal defect; Treatment; Collagen

Introduction

Cutis laxa type II (MIM 219200) is a rare congenital disorder first described by Debre et al. in 1937. During the period from 1937 to 2008, there have been 34 (25 females and nine males) well-documented published cases of this rare disorder. In 2009, I reported the thirty fifth case of this disorder and I also reviewed the previously published thirty four well documented cases [1,2].

The disorder is characterized by characteristic dysmorphic facial features and the unique association of generalized cutis laxa (loose skin from birth with redundant folds, generally sparing the face), and developmental delay. Consanguinity of

the parents is commonly observed. An autosomal recessive inheritance has been suggested [1-6].

Case Report

A dysmorphic girl (**Figure 1**) was referred at the age of four years (early April 2016) because of skin abnormality and delayed speech. Her growth was adequate (height, 103 cm; weight, 23 kg).

The girl had the characteristic phenotypic manifestation of type II recessive cutis laxa (Debre type) with redundant loose skin present since birth. This skin abnormality was evident on the limbs and abdomen.

She was born as the third child of healthy consanguineous parents of Arabian origin, who were cousins. She has healthy older sister aged six years, and health younger brother aged 9 months. There was no history of similar illness in any relative.

The motor development was acceptable, but her language was poorly developed. She was saying no meaningful words. However, the parents considered many of her words understandable to them and they were able to communicate with her. Examination of the cardiovascular system was normal except for the presence of a systolic murmur. Echocardiography revealed the presence of a small atrial septal defect (**Figure 2**).



Figure 1 A dysmorphic girl with cutis laxa (Debre type).



Figure 2 Echocardiography image showing evidence of small atrial septal defect.

Simple tests of mental abilities revealed her ability of drawing a circle and a square after being demonstrated by her father. Brain MRI revealed normal findings. Dysmorphic facial feature included, broad and depressed nasal bridge, and the eyes were wide set with down-slanting palpebral fissures. **Table 1** summarizes the presenting features.

Table 1 Presenting features.

Consanguinity
Delayed speech with poor language development
Hypertelorism
Depressed flat nasal bridge
High palate
Cutis laxa
Small atrial septal defect

The decision was made to treat her with skin dietary supplement in an attempt to improve her skin abnormality. Treatment was approved by the scientific committee of Iraq headquarter of Copernicus Scientists international panel. She was given collagen supplement in form of capsules containing 1g collagen. The capsules was to be given on alternate days. The parents were mixing the powder from the capsules with the broth she was eating at lunch.

After two months of collagen supplementation, a significant improvement occurred in the skin abnormality with significant reduction in the redundant skin without observing any unwanted effects.

The parents thought that the improvement in her condition was associated with improvement in her mood and enhanced her attempts to communicate and play with other children. The parents opinion was considered highly valid as it was very uncooperative with examination before treatment and was refusing talking photos and was running away and hiding her face when attempting to photo her. After treatment, she was much more cooperative and accepted taking photos without any resistant.

Discussion

The classical form which was originally described by Debre et al. in 1937, is also associated with pre-and postnatal growth retardation, and skeletal abnormalities including late closure of the fontanel, dislocation of one hip. This case seems to have rather milder disease with normal growth, and no obvious skeletal abnormality [1-6].

Fitzsimmons et al. have already suggested variable clinical presentation of this disorder 6. The occurrence of cardiac defect has not been reported before. However, this patient has atrial septal defect.

Most cases have been reported in Europeans, Turks, and Arabs. This patient is the 36th patient with this disorder and the second case in Iraq [1-6].

Conclusion

In this paper, I am reporting the 36th case of Cutis laxa (Debre type) which is the first case to be associated with atrial septal defect.

References

1. Debre R, Marie J, Seringe P (1937) Cutis laxa avec dystrophies osseuses. Bull Mem Soc Med Hop Paris 53: 1038-1039.
2. Al-Mosawi AJ (2009) The Thirty fifth case of Cutis laxa type II (Debre type). Int J Dermatol 48: 755-757.
3. Fittke H (1942) Ueber eine ungewoehnliche Form "multipler Erbabartung" (Chalodermie und Dysostose). Z Kinderheilk 63: 510-523.
4. Theopold W, Wildhack R (1951) Dermatocalasis in Rahmen multipler Abartungen. Mschr Kinderheilk 99: 213-218.
5. Reisner SH, Seelenfreund M, Ben-Bassat M (1971) Cutis laxa associated with severe intrauterine growth retardation and congenital dislocation of the hip. Acta Paediatr Scand 60: 357-360.
6. Fitzsimmons JS, Fitzsimmons EM, Guibert PR, Zaldua V, Dodd KL (1985) Variable clinical presentation of cutis laxa. Clin Genet 28: 284-295.