Electronic Physician (ISSN: 2008-5842) http://www.ephysician.ir
October 2015, Volume: 7, Issue: 6, Pages: 1391-1393, DOI: 10.14661/1391

Congenital Cutis Laxa Type 2 Associated With Recurrent Aspiration Pneumonia and Growth Delay: Case Report

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Type of article: Case report

Abstract
Cutis laxa is a connective tissue disorder caused by deficiency of fibro elastic plexus, which can involve multiple organs. It is inherited in autosomal dominant, autosomal recessive, and X-linked. Autosomal recessive cutis laxa type 2, which appears to compromise a spectrum of disorders, starts with severe wrinkly skin syndrome and leads to more severe diseases related to growth and developmental delays and skeletal anomalies. The clinical manifestations in some of cases of Cutis laxa consist of redundant loose skin, pre-and post-natal growth deficiency, mental retardation, large fontanels, and dislocation of the hips. The authors present the case of a female patient with involved internal organ disorder and delay in growth in addition to skin laxity in which gene sequence analysis of PYCR1 indicated C.797G>A mutation.
Keywords: connective tissue disorder, Cutis laxa Autosomal Recessive, wrinkled skin

1. Introduction

Cutis laxa is a connective tissue disorder caused by the deficiency of fibroelastic plexus, which can involve multiple organs, especially the skin (1). The prevalence of all types of Cutis laxa is unknown, but approximately 200 cases have been reported in the literature to date (2-4). In this disorder, the skin may be stretched easily from base tissue, and it returns to the initial status very slowly. It is inherited in autosomal dominant, autosomal recessive, and X-linked pattern. Also acquired forms of the disease have been reported. Inherited cases are often symptomatic at birth and present with involvement of the internal organs, delayed puberty, pulmonary emphysema, umbilical hernia, gastrointestinal diverticulitis, and rectal and uterovaginal prolapse (3-6). In this paper, we introduced a case of congenital Cutis laxa type 2 presented with skin hyperlaxity, involvement of the abdominal organs, and delayed growth.

2. Case presentation

2.1. Clinical presentation

A five-month old girl was admitted to Koodakan Hospital in Bandar Abass, Iran, with recurrent pneumonia and growth retardation. She was referred for the diagnosis and treatment processes. The parents reported similar frequent admissions for the same cause since birth. Various symptoms appeared after breast feeding, such as cyanosis, tachypnea, and coughing, and they disappeared after suctioning applied by the mother.

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Received: September 15, 2014, Accepted: August 17, 2015, Published: October 19, 2015
iThenticate screening: August 07, 2015, English editing: September 15, 2015, Quality control: October 01, 2015

2.2. History

The patient was the second child of the mother and was delivered by cesarean section at 31 weeks of gestation due to fetal bradycardia and placenta abruption. The infant weighed one kilogram when she was born, and she was hospitalized in the NICU for 56 days. The abdominal viscera were formed at birth, but they were not well developed and had ambiguous genitalia. The intra-abdominal viscera developed gradually. She was discharged after gaining 1.8 kilograms. During this time, she experienced a decrease in hemoglobin and was given blood. Because of the long-term oxygen dependency, her eyes and heart were examined to determine if they were healthy. The results of eye and heart examinations were normal.

2.3. Physical Examination

On physical examination, hypotonia, syndromic appearance, large head, flaccid body, loose skin, deformity of both wrists, rising palm lines, bulging vessels near the surface of the skin, and abnormal genitalia were apparent. Her cardiopulmonary examination was normal with no murmur. Episodes of apnea caused bradycardia and cyanosis. Rheumatologic examination showed hyper lax and dislocated joints.

2.4. Imaging

Echocardiography showed normal pulmonary pressure. Electromyography (EMG) showed signs of the myopathic motor unit's activities; there was normal motor and sensory nerve conduction with no evidence of peripheral neuropathies. The results of metabolic and immunologic tests were normal. An MRI indicated that she had a normal brain. A CT scan showed evidences of aspiration pneumonia. Percutaneous endoscopic gastrostomy (PEG) was placed to prevent aspiration pneumonia.

2.5. Family History

The patient's parents were consanguineously related, and they had undergone routine pre-gestational tests for genetic screening that showed normal ranges with no family history of genetic diseases. They refused to give their consent for the assessment of genetic diseases, and they insisted that their child was healthy. The patient was diagnosed with growth delay.

2.6. Diagnosis and Follow-Up

Ehlers Danlos syndrome was suspected in the patient due to skin laxity and rupture of the vessels after intravenous catheter insertion. However, Ehlers Danlos syndrome was ruled out because the skeletal and skin examinations were normal. In this syndrome, there is skeletal involvement, and the baby's musculoskeletal system was normal. In order to obtain a definitive diagnosis, blood samples were collected from the parents, their first child, and the baby, and a biopsy of the baby's skin was taken for genetic tests. The gene sequence analysis of PYCR1 revealed C.797G>A mutation, and cutis laxa type 2 was diagnosed. Eighteen months after she was born, she was hospitalized for aspiration pneumonia and treated for seven days. She died due to decrease in level of consciousness and renal failure.

2.7. Ethics of Case Report

Informed consent was obtained directly and in writing from the patient's parents for the publication of this manuscript.

3. Discussion

Cutis laxa syndrome is a rare disorder that occurs due to a deficiency in the production of elastic fibers and their destruction; the disorder can be hereditary or acquired. Cutis laxa type 2 is marked by wrinkled, inelastic skin, especially on the dorsal acral surfaces and abdomen, dislocation of the hips, intrauterine and postnatal retardation of growth, delayed development, and triangular dysmorphic facies with progeroid appearance, bulbous nose, prognathism, hypotelorism, epicanthal folds, blue sclera, large ears, and microcephaly (7-9). Mixed Ehlers-Danlos syndrome was rejected because there is a skeletal involvement in this syndrome, and the musculoskeletal system in this baby's case was normal. In order to obtain a definitive diagnosis, blood samples were collected from the parents, their first child, and the baby. Subsequently, a skin biopsy was taken for genetic tests, and Cutis laxa type 2 was detected. Although wrinkly skin syndrome appears to be more frequent than ARCL type II in clinical practice, this was not observed in our patient.

Our case was diagnosed by clinical examination, and it was confirmed molecularly by the identification of the C.797G>A mutation. Our patient presented with symptoms of hypotonia, syndromic appearance, large head, flaccid body, and loose skin which was different from previously reported (10-12). Our patient had no sign of hernia, but some studies have reported hernias in such patients (10, 12). Our case presented with hip dislocation, intrauterine and postnatal growth retardation, developmental delay, blue sclera, and large ears, but she had no characteristics of wrinkled skin, and flaccid body. The involvement of the lung and the stomach was noticeable in this case.

4. Conclusions

Cutis laxa syndrome is similar to many diseases, such as Ehlers-Danlos syndrome and Pseudoxanthoma Elasticum, so it is a difficult task to differentiate it from those. Skin biopsy may help differentiate it from other diseases, which, in turn, can lead to better diagnosis and, consequently, faster treatment. In the previous years when genetic analysis was not available, physical examination, skin biopsy and pathologic study were the bases for diagnosis, but, now, definitive diagnoses require genetic study.

Acknowledgments:

This research received no specific grant from the government, any public funding agency, or any non-profit organization.

Conflict of Interest:

There is no conflict of interest to be declared.

Authors' contributions:

All authors contributed to this project and article equally. All authors read and approved the final manuscript.

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