CASE REPORT

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Surgical Management of Inguinal Hernia in a Newborn with ARCL2B

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ABSTRACT Cutis laxa, being a rare disease has many accompanying disorders depending on the subtypes. It affects connective tissue that supports the body's framework in which the underlying pathology is related with elastin fibers. Herein, we presented successful management of a 20-day-old male admitted due to a unilateral inguinal hernia, atypical physical findings at physical examination those suggesting connective tissue disorder and with a sister who had a suspected diagnosis of Ehler Danlos Syndrome. To our knowledge, this will be the first case that discusses the uneventful operational management of an autosomal recessive cutis laxa type 2B patient in the English literature.

Keywords: Cutis laxa, recessive; cutis laxa; hernia, inguinal

Cutis laxa is a rare disease affecting approximately 400 families in the world with an incidence of 1 in 1 million and is equally distributed in both sexes.[1] It may be congenital or acquired. It affects connective tissue that supports the body's framework which provides structure and strength to the muscles, joints, organs, and skin. The underlying pathology is related with elastin fibers. It results in skin manifestations such as the aged face, loose redundant skin, and reduced elasticity; and extracutaneous manifestations such as diverticula at the gastrointestinal system or urinary system, gastroesophageal reflux, diaphragmatic atonia or a hernia, umbilical or an inguinal hernia and hypoplasia of corpus callosum.[2-4] In the literature, there are many reports about cutis laxa and its comorbidities but none is related with per operative management of the patient. To our knowledge, this will be the first case that discusses the surgical management of an autosomal recessive cutis laxa (ARCL2B) patient in English literature.

CASE REPORT

A 20-day-old male infant admitted to the outpatient clinic with a left inguinal hernia. He was born 1960 g at 40th gestational week with spontaneous vaginal delivery. He was 2300 g prior to operation. He had healthy parents who had a consanguineous marriage. He had two sisters who were 5 and 13 years old. The older sister was suspected as Ehler Danlos Syndrome. She was previously operated 3 times due to a recurrent bilateral inguinal

hernia. The patient had aged appearance, large fontanel, prominent forehead, blue sclera, prominent bulbose nose, congenital hip dislocation, loose redundant skin and prominent veins at physical examination (Figure 1). He didn't have any respiratory or urinary tract infections. He also had a huge left inguinal hernia and suspicious right inguinal hernia (Figure 2). He was consulted with genetic department due to his atypical findings and suspicion of Ehler Danlos Syndrome at his sister. He found to have a homozygous mutation c.283 T>C, p.(Cys95Arg), exon 3 in the PYCR1 gene at his genetic workup and did not have any mutation in ALDH18A1 gene. Per the genetic results, he was diagnosed as cutis laxa autosomal recessive type 2B (ARCL2B). His parents were diagnosed as heterozygous for the disease. While no mutation was found at younger sister, the older sister was also diagnosed as ARCL2B. Laboratory examinations revealed nothing except anemia. There was grade 1 hydronephrosis at his right kidney at abdominal ultrasonography. Echocardiography and cranial magnetic resonance imaging (MRI) were normal. Intubation of the patient was not difficult. The operation was performed with skin crease incision. The skin was so fragile that when using retractors, it was torn as a paper. A 5/0 polyglactin was used to repair hernia sac and 4/0 polyglactin to repair the fascia. Both the fascia and hernia sac was so fragile that a Gore-Tex mesh was used to prevent recurrence. The skin was also sutured with 5/0 polyglactin subcutaneously. The right inguinal canal was controlled via left inguinal canal with 5mm optic camera and processus vaginalis was found open. The right inguinal hernia was also repaired, but without using a mesh. He was followed up at neonatal intensive care unit (NICU) postoperatively regarding the risk of diaphragmatic atonia. He was extubated at 4th post-operative hours. He discharged from NICU the next day and discharged from hospital the day after. The wound healing was normal postoperatively. There was no dehiscence or infection at the wound. He has been free of any recurrence for the last two years.

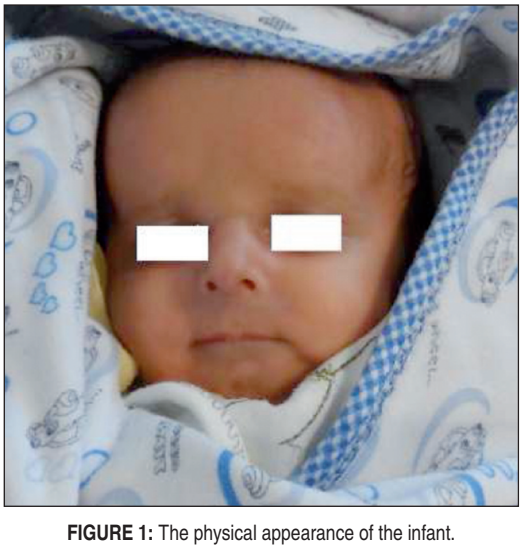

FIGURE 1: The physical appearance of the infant.

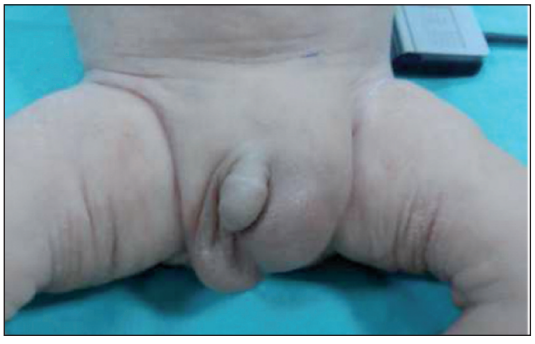

FIGURE 2: Appearance of the inguinal hernia.

DISCUSSION

Surgical management of patients with congenital connective tissue disorders requires intensive care. There is a huge improvement in diagnosis and management of connective tissue disorders in the last decade. Identification and clinical or molecular characterization of new phenotypes are the key factors for management of these diseases.[1]

Cutis laxa means lax or loose skin in Latin. It is also known as elastolysis which is one of the main representatives of this disorder. The clinical presentation and type of inheritance have a wide distribution. It has 20 subdivisions those may be acquired or hereditary; autosomal dominant, autosomal recessive or X-linked.[1] There are many genes those demonstrated to be affected; such as PC5S, PYCR1, ATPVOA2-CDG that causes an abnormal synthesis of elastin.

Autosomal Recessive Cutis Laxa type 2B (ARCL2B) is characterized by a mutation at pyrroline 5 carboxylate-reductase-1 gene (PYCR1, 1790 35.0001). ALDH18A1 gene is located on the long arm of chromosome 10 (10q24.1) and is related with the subtype of cutis laxa called De Barsy[5]. The skin symptoms of the ARCL2B subtype are more pronounced in the arms and legs. Children might have developmental or growth delays, failure to thrive, less intellectual capacity, joint laxity or skeletal malformations.

Inguinal hernia is one of the most common diseases in neonates. As the defect of the inguinal canal is narrower, the possibility of strangulation is higher which is an emergency in all age groups. Connective tissue disorders may increase the probability of an inguinal hernia by decreasing the durability of the inguinal canal.[5] The case presented here had a bilateral inguinal hernia with the defect at the right side is narrower than the left.

Atypical appearance and findings at the physical examination of the patient and the history that his sister had been operated 3 times due to recurrent inguinal hernia were alerting signs. His sister was suspected to have Ehler Danlos Syndrome. However, she did not have all components of the syndrome such as skin hyperelasticity. All other family members were normal. After genetic counseling, both of their findings were found to be consistent with ARCL2B. Possible accompanying disorders were investigated but he did not have any comorbidities. He was hemodynamically stable through the operation and no problem was encountered in regarding the anesthesia. Because patients with cutis laxa have been reported to have diaphragm atonia which could be a life-threatening issue while extubating, he was followed up at NICU postoperatively.

The major technical issue with the surgery was the reduced elasticity of the connective tissue. The durability of the tissues was incredibly decreased. Even handled in gentle and with care, retractors and needle tore the fascia and skin easily. Thinner sutures were needed to be used in order not to tear the tissues. Laparoscopy was performed via left inguinal canal to see if the right inguinal canal was open or not. It was necessary to perform laparoscopy; because patients with cutis laxa are more prone to bilateral inguinal hernia and he had a sister with recurrent bilateral inguinal hernia.[6,7] It also would be perceptive not to omit contralateral inguinal hernia and to prevent any unnecessary anesthesia.

There are very few cases those require mesh for treatment of an inguinal hernia. The defect at the left side was very wide and tissues were so fragile in the case presented here that a mesh was nee ded to be used. Mesh was used to increase the durability of the tissues. The defect at the right side was narrow enough to allow primary repair. Patients with cutis laxa are more prone to surgical infections due to the inability of the tissues to recover quickly. The mesh itself also increase the probability of the wound infection. However, the healing period of the infant was hassle-free.

Even the patient presented here have most pathognomonic features of ARCL2B such as aged appearance, large fontanel, prominent forehead, blue sclera, prominent bulbose nose, congenital hip dislocation, loose redundant skin and prominent veins; the operative management was almost uneventful. He was treated for congenital hip dislocation one month after the operation and hydro nephrosis did not increase.

The case is presentative for the uneventful operational management of ARCL2B patient when sufficient precautions are taken. To our knowledge, mesh repair in a patient with congenital connective tissue disorder would be a better and safer option.

Informed Consent

Written and verbal consent were taken from the parents.

Source of Finance

During this study, no financial or spiritual support was received neither from any pharmaceutical company that has a direct connection with the research subject, nor from a company that

provides or produces medical instruments and materials which may negatively affect the evaluation process of this study.

Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

All authors contributed equally while this study preparing.

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