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Barth syndrome-congenital skin aplasia (clinical case).

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Abstract

The presented clinical case refers to a female newborn who had various types of injuries since birth. The diagnosis was made by visual evaluation - congenital aplasia type VI - Barth's syndrome (BS), although all studies were done and consultations with doctors of various specialties were carried out. Based on the condition, a minimal intervention was decided, in order to avoid complications/infection, a conservative treatment was prescribed, with antibacterial ointments and a wet antibacterial gauze bandage. The desired result was achieved very soon, the skin began to regenerate and as a one-year monitoring showed, the limb was fully rehabilitated.

Keywords: Barth syndrome, congenital skin aplasia, treatment

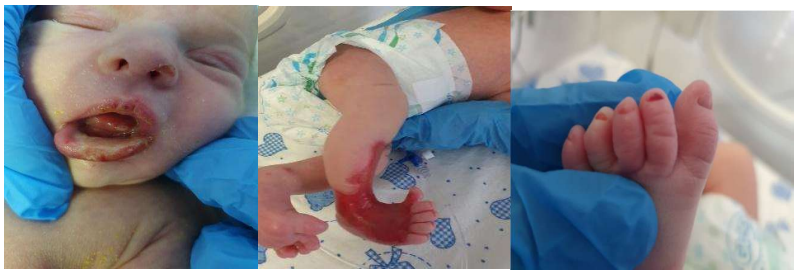
Barth syndrome, initially classified by Frieden in 1989 [3], encompasses a spectrum of dermatological conditions categorized into six groups, with the sixth group specifically delineating the features of the syndrome [4]. Barth syndrome, also referred to as Congenital Localized Aplasia of the Skin (CLAS) and Bullous Epidermolysis (BE), is characterized by congenital absence of skin and is classified as a subtype of epidermolysis bullosa.

Barth syndrome (BS) is typified by a triad of clinical features: congenital aplasia of the skin (ACC), epidermolysis bullosa (EB), and potential nail abnormalities including congenital absence, nail dystrophy, or subsequent loss. Additional associated congenital anomalies may include pyloric atresia, ureteral stenosis, renal pathologies, underdeveloped ear canal, flattened or broadened nasal bridge, and widely spaced eyes.

The objective of this study was to determine an effective treatment regimen for a newborn diagnosed with Barth syndrome, aiming to achieve wound resolution with minimal intervention and promote skin regeneration.

Materials and Methods: A female newborn was admitted to the Iashvili Children's Republican Clinic on the fifth day postpartum, presenting with a congenital skin defect characterized by the absence of the epidermal layer on the right leg, specifically on the dorsal surface of the tibia and ankle. The lesion exhibited well-defined borders and was covered with a red, thin, translucent membrane. Additionally, the newborn displayed a bullous, eroded rash on the mucous membrane surrounding the lips and on the wrist joint. There were no consanguineous relationships among the parents, and no history of skin or connective tissue disorders or autoimmune diseases was reported. The newborn girl was the product of the second pregnancy, with the first ending in artificial abortion, indicating a history of multiple pregnancies. The mother was 20 years old, and the father was 29 years old. Prenatal care was provided to the mother, and the pregnancy was uneventful. The girl was delivered via cesarean section at 39 weeks gestational age, with a birth weight of 3000 grams and no signs of asphyxia. Apgar scores were 7/8 at birth. Umbilical venous catheterization was performed. The newborn was conscious, exhibiting forced posture and altered expression, with a hyperergic response to stimuli and moaning vocalizations. Auscultation revealed equal breath sounds bilaterally. Abdominal examination was unremarkable, with no tenderness on palpation. Normal female genitalia were observed, and dysmorphic features were absent. Oxygen saturation was 96% on room air, with an FiO2 of 21%, and the body temperature was 36.6°C. The clinical condition was carefully evaluated, and the newborn was placed under a radiant warmer and cardiorespiratory monitoring. The skin defect was managed with sterile antibacterial bandaging, and the eroded rash was treated with Betadine solution and antibacterial ointments.

Before treatment:



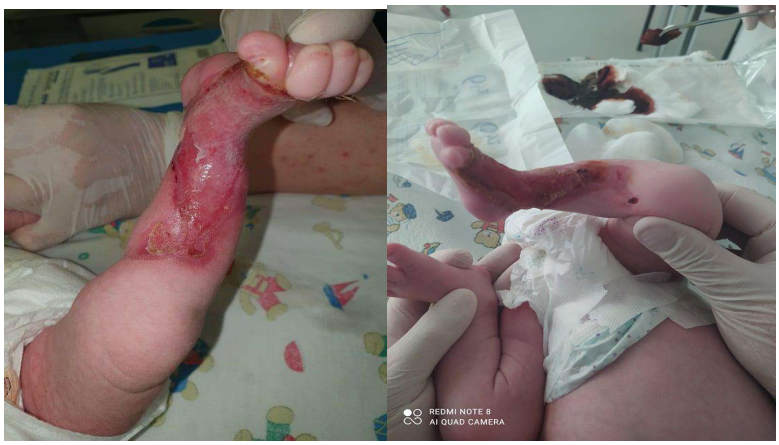
Study procedures included complete blood count (CBC), basic metabolic panel (BMP), venous blood gas (VBG), blood culture, lactate assessment, liver function tests (LFT), renal function tests (RFT), X-ray imaging, head ultrasound (HUS), abdominal ultrasound (AUS), and echocardiography (ECHO). Laboratory findings revealed total bilirubin (T-BIL) levels at 170 μmol/L and C-reactive protein (CRP) levels at 6 mg/L. Liver function tests demonstrated values within normal limits, with urea levels at 22.59 mmol/L and creatinine levels at 0.54 mg/dL.

The patient underwent consultations with dermatologists, ophthalmologists, and burn specialists, as well as a geneticist for genetic analysis, specifically KRT5 and KRT14 gene sequencing, which was recommended to confirm the diagnosis of congenital skin aplasia type VI, also known as Barth Syndrome. Diagnosis was primarily clinical, based on distinctive signs and symptoms of the condition, while genetic analysis and histological skin examination were deemed necessary for

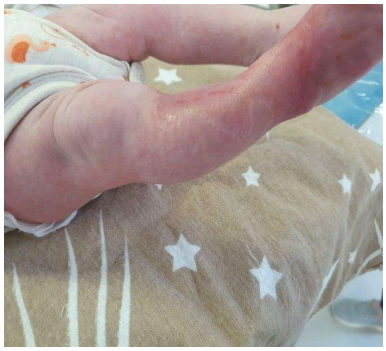
definitive confirmation. The mode of disease transmission was considered to be either autosomal dominant or sporadic, with the latter attributed to a genetic mutation, given the healthy status of both parents.

Management recommendations emphasized a simple and targeted approach to optimize wound healing outcomes, avoiding active surgical intervention to prevent infection and scarring. Symptomatic management was endorsed, while prophylactic antibacterial treatment with antibiotics was deemed unnecessary. Close monitoring and ongoing multidisciplinary care were recommended to ensure the patient's well-being and facilitate optimal recovery.

After treatment:



After 1 year:



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ბარტ სინდრომი (კლინიკური შემთხვევა)

თამარ ებანოიძე, გიორგი დურგლიშვილი, ლელა ბერიძე, ნატო დურგლიშვილი, ნინო გრძელიძე, სოფიო საფარიძე, მარიამ მერკვილაძე

კანისა და ვენ.სნეულებათა ს/კ ეროვნული ცენტრი; .ბათუმის რესპუბლიკური კლინიკური საავადმყოფო; სს საქართველოს კლინიკები

აბსტრაქტი

წარმოდგენილი კლინიკური შემთხვევა ეხება ახალშობილ გოგონას, რომელსაც დაბადებიდანვე სხვადასხვა ტიპის დაზიანებები აღენიშნებოდა. დიაგნოზი დაისვა ვიზუალური შეფასების საფუძველზე - კანის თანდაყოლილი აპლაზია VI ტიპი - ბარტ სინდრომი (BS), პაციენტს ჩაუტარდა ყველა საჭირო კვლევა და კონსულტაცია სხვადასხვა სპეციალობის ექიმებთან. მდგომარეობის საფუძველზე მიღებულ იქნა მინიმალური ჩარევის გადაწყვეტილება, რათა თავიდან აცილებულიყო გართულებები, ისეთი როგორიცაა ინფექცია. დაინიშნა კონსერვატიული მკურნალობა, ანტიბაქტერიული მალამოებითა და სველი ანტიბაქტერიული სახვევით. სასურველი შედეგი ძალიან მალე დადგა, კანის მთლიანობამ ნელ-ნელა დაიწყო აღდგენა და ერთწლიანი მონიტორინგის შედეგად დადგინდა, რომ კიდური სრულად რეაბილიტირდა.

საკვანძო სიტყვები : ბარტ სინდრომი, კანის თანდაყოლილი აპლაზია, მკურნალობა