Case report on: - A case report on management and outcomes of Bart Syndrome

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A limited number of (neutropenia), a weaker and enlarged heart, skeletal myopathy, recurrent infections, and short height are all features of the rare illness known as Bart syndrome. Bart syndrome nearly always affects men. Dilated cardiomyopathy frequently manifests at birth or during the first few months of life in males with Bart syndrome. The heart muscle weakens and loses its capacity to pump blood with time. Elastic fibres may replace muscle fibres in some parts of the heart muscle in people with Bart syndrome, which adds to cardiomyopathy. Endocardial fibroelastosis is the name of the disorder that causes the muscle to thicken and affects how well it pumps blood. Heart failure can result from heart issues in persons with Bart syndrome. Rarely the cardiomyopathy improves with time, and those affected eventually show no signs of cardiac illness. Main symptoms and significant findings: A 5 months old baby boy was admitted to the hospital, Wardha on dated Jun 23 2022. Chief complaints of enlarged heart, low blood cell count, weakness of muscles, and fatigue. Additionally, there can be increased levels of chemicals like 3-methylglutaconic acid and 2-ethyl hydracrylic acid in the urine or blood. The primary diagnosis, therapeutic interventions, and outcomes: The patient was treated conservatively without surgery after the doctor discovered Bart syndrome following a physical examination and investigation. When the patient was brought to our hospital, the wounds had crusts and dead tissue but showed no evidence of infection. We removed crusts and dead tissue using mechanical and hydrodebridement techniques with sterile saline. Next, we administered an antimicrobial ointment to prevent disease and encourage wound contraction. A gauze bandage infused with petrolatum was also used to keep wounds moisturized. Antibiotics weren't administered intravenously or orally. The skin lesions were entirely epithelialized and healed after three months, leaving just a pigmented scar in their place. He underwent all treatments, and the results were positive. Her complaints have slightly decreased. Conclusion: We describe a case where Bart syndrome symptoms were found following our patient's enlarged heart, low blood cell count, muscle weakness, and exhaustion. These symptoms exactly matched those listed as official Bart syndrome symptoms. An uncommon congenital skin condition known as Bart syndrome has a distinctive clinical appearance. It's crucial to search for related oddities. The prognosis for the syndrome is generally good, however, the best results can be achieved by treating it as soon as feasible.

Keywords: Bart syndrome, hydro debridement, epithelialization, endocardial, fibroelastosis.

INTRODUCTION

Bart syndrome, also known as aplasia cutis congenital type VI, is a rare genetic condition marked by mucocutaneous blisters, missing or dystrophic nails, and congenitally localized skin loss. TAFAZZIN gene mutations bring on Barth syndrome. The tafazzin protein is made using instructions from the TAFAZZIN gene. This protein is found in the mitochondria, or energy-producing parts of cells. Cardiolipin, a lipid with essential functions in the mitochondrial inner membrane, is modified by the tafazzin protein. After being modified by tafazzin, cardiolipin is necessary for preserving the mitochondria's integrity, carrying cellular proteins, and producing energy. The Tafazzin proteins produced by TAFAZZIN gene mutations either have very little or no function. As a result, Tafazzin cannot change cardiolipin, and levels of functional cardiolipin are decreased. Additionally, for unknown reasons, a cardiolipin variant known as monolysocardiolipin (MLCL) develops. An excess of MLCL and a shortage of functional cardiolipin undermine healthy mitochondrial structure and functioning. Tissues with high energy requirements, such as the heart and skeletal muscles, are susceptible to cell death because mitochondrial energy generation is constrained. The affected individuals' white blood cells also have abnormally shaped mitochondria, which could affect how well they grow, mature, and work, leading to neutropenia. Other Bart syndrome signs and symptoms are likely brought on by malfunctioning mitochondria.
Patient-specific information: A 5 months old baby was admitted to A.V.B.R. Hospital on dated Jun 23 2022. Chief complaints of enlarged heart, low blood cell count, weakness of muscles, and fatigue. After physical examination and investigation physician diagnosed a case of Bart syndrome. Baby born with full-term delivery with cephalic presentation.

The primary concern and symptoms of the patient:

The main symptoms were an enlarged heart, low blood cell count, muscle weakness, and exhaustion. At the time of admission, these were the main symptoms that were seen.

Medical, family, and psychological history:

There is no history of surgery in the current case. There are four people in his nuclear family, which he belongs to. Except for the patient, who appears to be quite fatigued, all family members were in good health. Following medical management, she had a positive outcome.

Relevant past intervention with outcomes:

Since birth, the patient has had a history of Bart syndrome; for additional care, they were hospitalized in Chandrapur. After being diagnosed with Bart syndrome, she got a positive outcome following several specific tests and examinations.

Clinical finding:

The patient is conscious and well-oriented to she is relative, and she is all motor responses. They were active. She is body guilt moderate. She is weight is 5kg. Vital parameters are normal. She is milestones and development are normal.

Timeline:

21 days ago, she was admitted to the hospital for treatment of Bart syndrome. The physician prescribed the drug INJ. M.V.B.C. (Multivitamin b Complex) once a day INJ. NEOMOL when needed INJ.PAUSE (TRANEXAMIC ACID INJECTION) Some supplementary milk was given to enhance immune function.

Diagnostic assessment:

Based on the patient's history, Physical examination, abdominal palpation, and some specific investigation, Bart syndrome was observed. In Angiography, enlargement of the pericardium was observed. His motor responses, milestone, and development were normal no evidence of faecal body lesion. On physical examination, symmetrical well demarcedated erosions over the anterior medial aspect of both lower limbs starting from the ankle and extending to the dorsal and lateral plantar parts of the feet suggestive of cutis aplasia. In addition, she had blisters on both upper and lower limbs. Later, she developed blistering lesions on the trunk in response to minor trauma are friction suggestive of epidermolysis Bullosa. There was no nail or scalp involvement. The rest of the is physical examination was routine.

Diagnostic challenges:

There are no diagnostic challenges faced during she is treatment.

Diagnosis:

After a physical examination and some specific investigations, the pediatric physician diagnosed the case with Bart syndrome.

Therapeutic interventions:

Medical management was provided to the patient. INJ. MVBC (Multivitamin b Complex) once a day INJ. NEOMOL when needed INJ.PAUSE (TRANEXAMIC ACID INJECTION) INJ. LEVIPRIL twice in a day INJ DEXA once in a day
nebulization with dealing and budecort were given twice a day. Some supplementary milk was given to enhance immune functions.

Follow up and outcomes:

Pediatric physicians and surgeons were taken follow-ups daily. And advice for taking all treatments for a good outcome.

Adverse and unanticipated event: there was no any Adverse and unpredictable event during medical treatment.

Discussion:

Bart syndrome was first described in 1966 by Therapeutic Bart, who published 26 family members with the affected condition. BS consists of a tried epidermolysis bullosa, aplasia cutis, and male dystrophy. However, there can be cases without nail involvement. Like the index case, the inheritance pattern of BS is mainly autosomal dominant, with few reported cases of new mutations. This baby belongs to the sporadic category due to the absence of family history. In addition to mucocutaneous involvement, BS has other associations such as pyloric atresia, microtia, flat nasal bridge, and hypertelorism. None of these abnormalities was there in this baby. The diagnosis of Bart syndrome is usually made on clinical features, but the microscopic appearance of the affected skin may aid the diagnosis. The management of Bart syndrome is mainly based on supportive care.3 Local application of diluted povidone-iodine, fusidic acid, and application of non-adhesive bandages impregnated with dexpanthenol and chlorhexidine are recommended forms of wound care and it is not prophylactically. The prognosis of Bart syndrome is considered to be good, and they have an average life expectancy. However, it is essential to protect these children from hypothermia, infections, and excessive friction. 4-21

In a family with EB simplex and congenital skinlessness, four generations were affected, according to Kanzler et al. According to them, the family mentioned by Bart et al. most likely also had widespread EB simplex of the Koebner type. They performed the first electron microscopy and immunofluorescent mapping on their patient's congenitally absent skin and regions of skin fragility. The results from both the immunofluorescence and electron microscopy were similar. The region with congenital skin deficiency was shown to have the same pathogenic alterations as EB, according to these ultrastructural results. They noted that congenital skin deficiency was linked to all of the main hereditary forms of EB. They claim that aplasia cutis congenital is not a distinct entity but a clinical indication of EB in utero, as is a localized loss of skin.22-30

Conclusion:

A uncommon congenital skin condition known as Bart syndrome has a distinctive clinical appearance. It's crucial to search for related oddities. The prognosis for the syndrome is generally good. However, treating it as soon as feasible can achieve the best results. Without sophisticated interventions, management can be conservative and rely on a straightforward technique for quick and ideal healing. Close It was advised that the patient be monitored.

REFERENCES
