Bart Syndrome: A Case Report of Neonatal Disorder

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Introduction:

A medical condition characterized by the congenital absence of skin, predominantly affecting the lower extremities, blisters on both skin and mucous membranes, and congenital absence and deformities in nails, has been identified within a familial context(1). The inheritance pattern aligns with a fully penetrant, autosomal dominant gene(1).

In 1966, Bart introduced Bart syndrome, a condition marked by the simultaneous occurrence of congenital epidermolysis bullosa, localized congenital absence of skin affecting the extremities, and the abnormal shedding or dystrophy of nails(2). This syndrome is fascinating in the medical field due to its more optimistic prognosis when contrasted with other epidermolysis bullosa types(2). This type is known as Aplasia cutis congenital type VI(3).

In this study, we have presented a case of a neonate with some disorders that guide us to different diagnoses and syndromes, such as CHIME syndrome and Barth syndrome, that have contributed.

Case presentation:

In October 2023, a premature baby was born at Ganjavian Hospital of Dezfull. His arrival was not without complexities, as he brought a puzzling array of skin symptoms and multi-organ anomalies. This baby was born after 33 weeks of pregnancy to a 24-year-old Iranian mother experiencing her first pregnancy. The hospital's gynecological surgery team performed the cesarean delivery. As the baby took his first breaths, the attending medical staff noted an initial Apgar score of 8/10, which swiftly improved to a reassuring 9/10 within the first five minutes(Table 3).

Within the context of the baby's family history, a significant detail emerged—the parents were closely related by blood, and there were no known instances of similar anomalies within their respective families. The decision to opt for a cesarean section was motivated by the presence of meconium in the baby's amniotic fluid, a precautionary measure taken to protect his well-being. Regarding physical stature, both parents fell comfortably within the normal range—standing tall at approximately 180 centimeters with a weight of 80 kilograms for the father and a height of 168 centimeters with a weight of 64 kilograms for the mother. Furthermore, the mother's blood type was identified as A+.

Upon his arrival, the baby weighed a delicate 1500 gr and measured 42 cm in length, with a head circumference of 31.5 cm. The temperature at birth held steady at 36.7C while vital signs were meticulously recorded—his heart rhythm was 140 beats per minute, and his respiratory rate was 68 breaths per minute. From the moment of birth until the time of this composition, spanning approximately four days, the baby has been under the dedicated care of the Neonatal Intensive Care Unit (NICU). During this period, he has displayed pronounced intercostal retractions, the poignant sound of grunting, and the telltale sign of nasal flaring. A thorough examination at birth also unveiled an open rectum, and apnea episodes further added to the complexity of his condition.

Despite these challenges, it is noteworthy that the baby was not macrosomic, nor did he suffer from intrauterine growth restriction (IUGR). Hearteningly, there were no indications of deceleration or irregular heart rate patterns, and premature rupture of membranes (PROM) did not factor into this medical narrative.

As our gaze approaches the baby's delicate skin, we encounter a mosaic of anomalies—ecchymosis, skin breakdown, and even the absence of skin formation in specific areas. These areas encompass the precious head, expressive face, tender neck, left hand, dainty feet, and even the tiny genitalia. The baby's left earlobe reveals aplasia and an entire formation of the right ear is regrettably absent. Additionally, aplasia extends into the nasal region, further underscoring the intricate nature of this medical tapestry(figure 1).

During the initial assessment of the baby, we encountered a concerning array of symptoms related to their eyes. The corneal cloudiness and the strikingly white appearance of the pupils immediately caught our attention, pointing to the presence of corneal opacities and an unusually smooth corneal surface. Recognizing the gravity of this situation, we promptly requested a consultation with an ophthalmologist to evaluate the baby's ocular health thoroughly.

The subsequent eye examination confirmed the presence of corneal cloudiness, but it was intriguingly limited to the central cornea, with the periphery maintaining a reassuringly normal appearance. This finding led the medical team to conclude that the corneal opacity fell within an acceptable range of variation. It is worth noting that the baby displayed an unusual inability to keep their eyelids open, a phenomenon often associated with underlying issues. However, notably absent was any sign of ectropion, the outward turning of the eyelids, which was a positive indication.

Further investigations considered the presence of clubbing, the thickening of the fingertips, initially observed during the baby's examination. After careful consideration and diagnostic assessment, the possibility of KID syndrome was introduced. In response to this potential diagnosis, a treatment regimen was established, comprising the administration of erythromycin eye ointment twice daily to address the corneal issues and a weekly application of lubricating ointment to alleviate ocular discomfort.

In dermatological evaluations, our scrutiny extended to the baby's delicate skin. The findings revealed disseminated vesicles and skin ecchymoses, forming a pattern indicative of dermatolysis. During this examination, an alarming discovery was made—a lack of reflex to light, an observation that raised concerns about possible visual impairment, particularly in light of the ear anomalies in the baby's medical profile(figure 4).

The baby presented us with several additional challenges in the broader context of medical examinations. Notably, we could not detect audible heart sounds during our assessment, and when they were faintly present, it raised potential red flags regarding cardiac issues. Additionally, the baby exhibited bilateral cryptorchidism, a condition characterized by undescended testes, and we observed early separation of the umbilical cord, which warranted close monitoring. Encouragingly, the results of the blood culture and C-reactive protein (CRP) levels fell within the normal range, offering a glimmer of reassurance amid this complex medical journey(Table1,2).

Table 1: laboratory findings

Laboratory

BS: 73	MCH:37.14	RBC:4.47	MCV: 106.4	Lymph:51.3	NRBC:70
Ca:8.8	MCHC:35.2	HB:16.6	Poly:40	MPV:9.3	Mono:5.5
WBC:11.6	RDW:11	Hct:47.4	EOS:3	Plt:319	

Table 2: ABG findings

ABG of the umbilical cord

pH: 7.374	pO2:34.9

pCO2:45.3	Hct49
After three h	**After three h**
pH:7.29	pO2:55.2
pCO2:37.5	

Table 3: APGAR table of patient

APGAR Score

20M	15M	10M	5M	1Minute	2	1	0	**sign**
2	2	2	1	1	Pink	Acrocyanotic	Pale	color
2	2	2	2	2	>100bpm	<100bpm	Absent	Heart Rate
2	2	2	2	2	Cry&active	Grimace	No response	Reflex irritability
2	2	2	2	1	Active motion	Some flection	Limp	Muscle tone
2	2	2	2	2	Good crying	Weak cry	Absent	Respiration

To manage the respiratory distress, skin issues, visual impairments, and mental challenges presented by the newborn., the decision was made to admit the baby to the Neonatal Intensive Care Unit (NICU) for specialized care(figure 2,3). The baby was placed in a temperature-controlled incubator within this unit to provide the optimal environment for their recovery and well-being.

A comprehensive regimen of medications and therapies was established for the baby's treatment. To alleviate the breathing difficulties, the baby was administered gentamicin and ampicillin intramuscularly to combat potential infections. The skin conditions were treated with a combination of topical medications, including an emulsion of RDen, mupirocin ointment, Lubratax ointment, erythromycin ointment, and gentamicin eye ointment. These treatments aim to alleviate skin issues and promote healing.

In addition to these measures, meticulous eye care was essential due to visual impairments. Gentamicin eye ointment and injections managed potential eye infections and protected the baby's delicate eyesight.

Furthermore, a portable brain ultrasound on the newborn revealed no evident signs of hydrocephalus or midline shift, providing some relief from concerns related to these conditions. There were no indications of intraventricular hemorrhage (IVH) or cerebral parenchymal lesions, underscoring the importance of early monitoring and intervention to safeguard the baby's neurological health. The multidisciplinary approach in the NICU, combining respiratory support, dermatological care, eye treatments, and neurological assessments, aimed to provide the newborn with the best possible care and enhance their prospects for a healthy future. Close monitoring and tailored interventions will continue to be essential to the baby's ongoing care and treatment plan.

Despite all efforts for further investigations and actions to treat the patient, the baby's family finally discharged the baby from the hospital with personal consent.

Discussion :

The mature epidermis is a stratified epithelial tissue composed predominantly of keratinocytes(4). A syndrome, as related to genetics, is a group of traits or Conditions that tend to occur together and Characterize a recognizable disease. Some syndromes have a genetic Cause.

Bart syndrome is an unusual condition that combines features from two distinct disorders, namely aplasia cutis congenita (ACC) and epidermolysis bullosa (EB)(5). ACC, a rare congenital anomaly characterized by the absence of skin, was initially documented in 1767 by Cordon(5). Frieden's classification system categorizes ACC into nine groups based on location and associated abnormalities(5).

Inherited EB is a group of genetically transmitted Skin disorders. Ep can be part of other syndromes(6). It can be marked by skin and mucous membrane fragility, resulting in blisters with minimal trauma(7). The clinical diagnosis of aplasia cutis congenital, specifically Bart syndrome, hinges primarily on identifying classical cutaneous manifestations, with the extent of involvement contingent upon the mode of inheritance(8).

In alignment with Omran et al. and Sharif, our case also exhibited skin lesions and ear malformation(3). In this case, we have seen standard Apgar scores like Sharif et al. (3). It could be better to have a skin biopsy and Brain MRI. To achieve a precise postnatal classification of inherited Epidermolysis Bullosa (EB), it is imperative to employ skin biopsy as a fundamental diagnostic tool(8). This biopsy specimen should undergo comprehensive examination, incorporating a blend of ultrastructural and antigenic assessments through transmission electron microscopy, immunofluorescence antigenic mapping, and investigations utilizing EB-specific monoclonal antibodies(8). In the treatment of this child, due to a skin disorder, the use of CPAP was not possible. Additionally, vascular access was not performed, and an umbilical catheter was placed. Antibiotics were administered due to suspicion of a skin infection with Staphylococcus aureus, and the treatment was carried out with ampicillin and amikacin.

Management of Bart syndrome involves a comprehensive approach, including conservative measures, secondary intention healing, and surgical interventions when warranted. Traditional care primarily focuses on localized wound treatment and infection control using systemic antibiotics(5). The routine administration of systemic antibiotics is not a standard practice, but they may be considered if there are concerns about infection. While conservative methods suffice for most patients, those with substantial or deep wounds may require surgical interventions such as skin grafting or local flap procedures(5).

Conclusion:

Bart syndrome, an infrequent congenital skin disorder characterized by its distinct clinical features, emphasizes prompt and conservative management's importance in optimizing outcomes. Vigilant patient monitoring is advised for tracking progress. *The first examination of newborns Can guide us to critical Situations. Some Signs are straight to the diagnosis, but others are conflicting, and one Should. Search for another disorder already in Syndromes, so physicians Should be aware of Many syndromes and associations so as not to miss the exact diagnosis.*

Keywords: Pediatric, Bart, Syndrome, Anomaly

Authorship List:

Author Contributions:

Author 1: Dr. Mohammad Amin Eqtedari: Author of the paper.

Author 2: Dr. Monireh Sharafi: Treating physician and provided scientific oversight for the manuscript.

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Conflict of Interest Statement:

The authors declare no conflicts of interest related to this research. No financial, personal, or professional affiliations or activities could be perceived as having influenced the study or the manuscript preparation. We are committed to ensuring the integrity and impartiality of our work.

Consent statement:

In this study, despite the unfavorable conditions experienced by the patient's parents and their pursuit of his referral to other cities for treatment, the patient remained in stable conditions and was hospitalized in the Neonatal Intensive Care Unit (NICU) due to the severity of the situation. Nevertheless, verbal and written consent was obtained from the parents for the academic publication of the material without disclosing identity and personal information.

Key Clinical Message:

Bart Syndrome, characterized by congenital skin absence, blistering, and nail abnormalities, presents complex neonatal challenges. This rare condition demands a multidisciplinary approach for accurate diagnosis and comprehensive care. Early recognition and management of associated anomalies are crucial, emphasizing the importance of thorough neonatal examinations.

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Images:

Figure 1: skin abnormalities and ear and nose abnormalities.

Figure 2: Skin Surface Erythemas

Figure 3: Separation and inadequate formation of scalp skin due to dryness and illness

Figure 4: Inappropriate conditions of the right ear

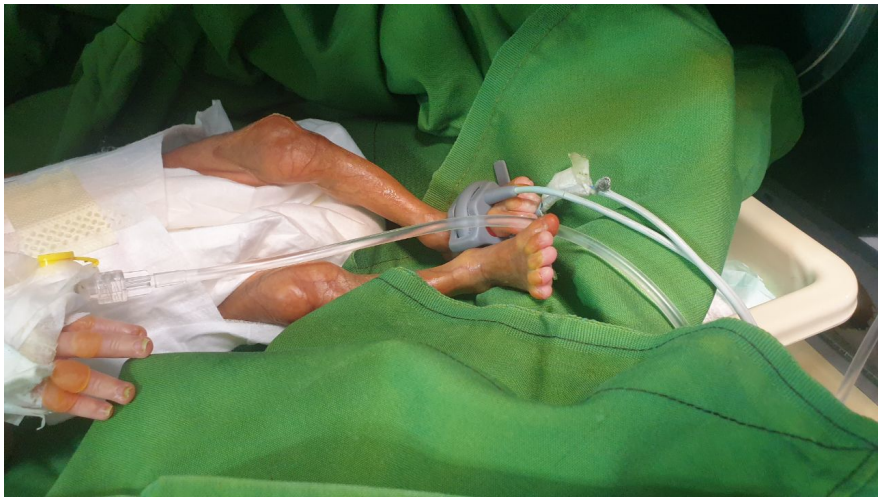

Figure 5: Dryness and inadequate formation of skin on the limb

Figure 6: Overall patient condition

Figure 7: patient X-RAY