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CASE REPORT

Lessons to Learn About the Misdiagnosis of a Rare Case in China: Bart Syndrome or Carmi Syndrome?

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Objective: We report a case of Carmi Syndrome in a neonate.

Aim: To share our lessons in diagnosis of the case of Carmi Syndrome.

Case Report: Carmi Syndrome is an extremely rare autosomal recessive genetic disorder characterized the coexistence of pyloric atresia and junctional epidermolysis bullosa, and with aplasia cutis congenita in approximately 28% patients. In this case, a full-term male neonate was born to a $G_4P_2^{+1}L^1$ multipara through cesarean section delivery in hospital in a non-consanguineous marriage with 4000mL of II°meconium-stained amniotic fluid. He was found extensive skin loss over lower legs and other parts, with scattered blisters and bilateral microtia. Plain abdominal X-ray revealed a large gastric air bubble with no gas distally. The mother had an intrauterine fetal loss previously for reasons unknown. The dermatologist diagnosed the newborn with Bart Syndrome, while the pediatric surgeon diagnosed congenital pyloric atresia(CPA). The parents refused further treatment and the neonate passed away about 30 hours after birth.

Outcome: The neonate passed away about 30 hours after birth.

Conclusion: Lessons from this case:①.Rule out Carmi Syndrome in patients with PA, and differentiate Bart syndrome and Carmi Syndrome in patients with abnormal skin manifestations. ②. For rare and/or severe diseases, multidisciplinary teams(MDTs) should be establish. ③. Genetic counseling and prenatal diagnosis are necessary prior to subsequent childbearings. ④.Termination of pregnancy might be contemplated if certain indicators are revealed.

Keywords: Carmi syndrome, bart syndrome, epidermolysis bullosa, EB, junctional epidermolysis bullosa, JEB, congenital pyloric atresia, CPA, aplasia cutis congenita, ACC, congenital localized absence of skin, CLAS, pyloric atresia, PA, epidermolysis bullosa with pyloric atresia, EB-PA, multidisciplinary teams, MDT

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Introduction

Carmi Syndrome(OMIM #226730) is an extremely rare autosomal recessive genetic disorder characterized the coexistence of pyloric atresia(PA)(OMIM #265950) and junctional epidermolysis bullosa(JEB)(OMIM #226650), and with aplasia cutis congenita (ACC)(OMIM #107600) in approximately 28% patients.[1] PA routinely presents non-bilious vomiting and feeding intolerance, while epidermolysis bullosa(EB) presents with blistering, skin erosions and ulceration in response to even slightest trauma, and ACC is characterized by localized or widespread absence of skin.

Swinburne and Kohler first described the association between EB and PA in 1967 and published the corresponding paper in 1968.[2] Carmi described 2 cases of ACC. The ACC of one case was extensive and was associated with PA and also EB-like disorder, and he contributed the genetic analysis.[3] A year later, the eponym "Carmi Syndrome" emerged in literature.[4] The incidences of EB and PA have been quoted as 19.6 per 1,000,000 live births[5] and 1 of 100,000 live births[6] respectively. The exact prevalence of the coexistence of PA-EB is unknown, but speculated to be[7] <1:1,000,000, and the carrier frequency has been estimated at less than 1/5000 live births.[8] About 100 cases have been documented worldwide to date.[1]

In terms of the molecular basis, its mutations often affect the laminin 332 genes, causing structural abnormalities in anchoring filaments of the lamina lucida and lamina densa, or disrupt hemidesmosomal proteins such as collagen XVII, integrin beta 4 (IGB4), integrin alpha 6 (IGA6), or plectin 1 (PLEC1).[1] Diagnosis of Carmi Syndrome is primarily clinical and usually straightforward, whereas the specific subtypes of EB and PA respectively require biopsy, light microscopy, direct immunofluorescence, transmission electron microscopy (TEM), intraoperative dissection, autopsy and so forth.

Due to the skin lesions, the affected patients are susceptible to sepsis, renal failure and other adverse clinical outcomes, and hence the disease carries a high mortality rate up to 75% with 50% of deaths occurring in the neonatal period. The median time of death was 30 days of life.[1] As a result, Carmi Syndrome is often fatal in neonates. Given the high fatality rate of Carmi Syndrome, it poses a challenge in treatment. In terms of EB, current management emphasizes wound care, pain control, and nutrition. Regenerative strategies, including stem cell and gene therapies, show promise in addressing defects and promoting healing.[6] While in terms of PA, excision of atretic pylorus and gastro-duodenostomy are suggested after the skin injury healed.

We herein delineate a case of Carmi Syndrome in a neonate, who was initially misdiagnosed as concomitant Bart Syndrome(OMIM #132000) and CPA one-sidedly respectively. This report has obtained written approval from the Ethics Committee of Chengdu Women's and Children's Central Hospital and consent from the legal guardian of the patient for images and other clinical information to be reported in the journal. The guardian understands that child's names and initials will not be published and due efforts will be made to conceal the identity

Case Presentation

A male neonate with a birth weight of 2800g, body length of 47cm, and Apgar scores of 9–10-10 was born via cesarean section delivery to a 33-year-old $G_4P_2^{+1}L_1$ mother at 38^{+5}-week gestation in hospital in a nonconsanguineous marriage with 4000mL of II° meconium-stained amniotic fluid in a tertiary first-class hospital in Chengdu, western China, in 2019.

The mother had a history of one intrauterine fetal loss at 7 months' gestation for undetermined cause, gave birth to a healthy male infant two years previously, and experienced a spontaneous abortion during her third pregnancy with specific details unavailable. This was her fourth pregnancy, and she underwent routine prenatal examination in the local county-level hospital during the mid-term of pregnancy. Given the history of previous stillbirths, the pregnant woman underwent comprehensive genetic screenings, including thalassemia gene screening, amniocentesis, and examinations of fetal chromosomal number chromosomal aneuploidy, and gene copy number variations at gestational age of 22^{+5} weeks at the most authoritative hospital in the province. However, all these investigations failed to reveal any abnormalities. At 35^{+6}-week gestation, the local hospital detected a serum alpha-fetoprotein (AFP) level in the pregnant women exceeding 1000 ng/mL. At 36^{+5}-week gestation, upon transferring to our hospital in preparation for delivery, ultrasound revealed mild polyhydramnios (Amniotic fluid index of 33.5 cm.)(Figure 1), chorioamniotic membrane separation(CMS), and an enlarged fetal gastric bubble (7.2cm×3.0cm) (Figure 2), and the pregnant woman was diagnosed with gestational diabetes (GDM) and suspected fetal digestive tract obstruction by the outpatient obstetrician.

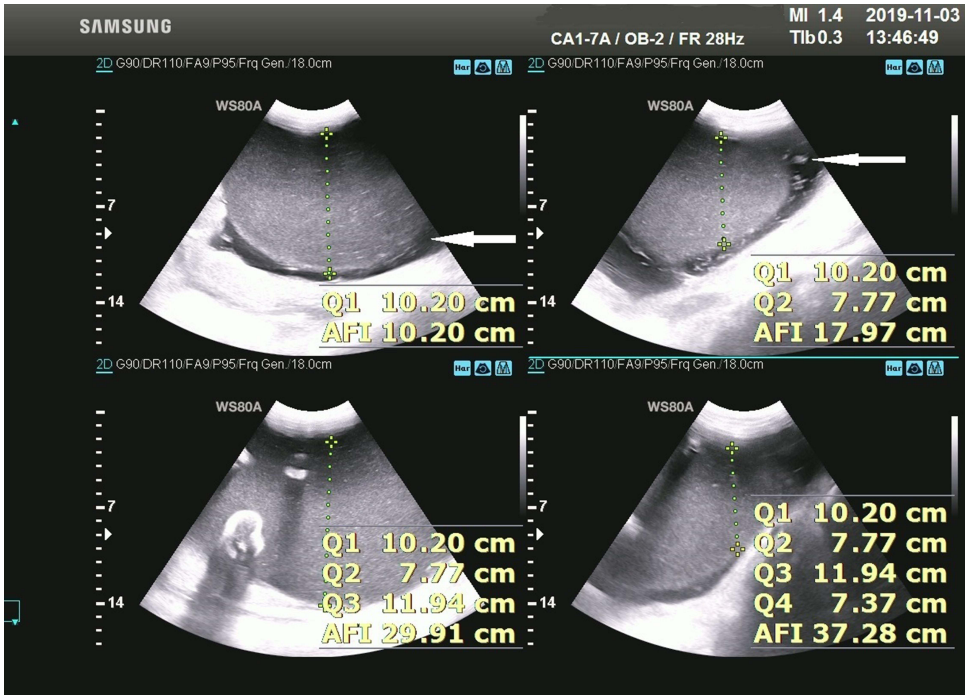

Figure 1 Ultrasonography shows mild polyhydramnios and also chorioamniotic membrane separation(CMS).

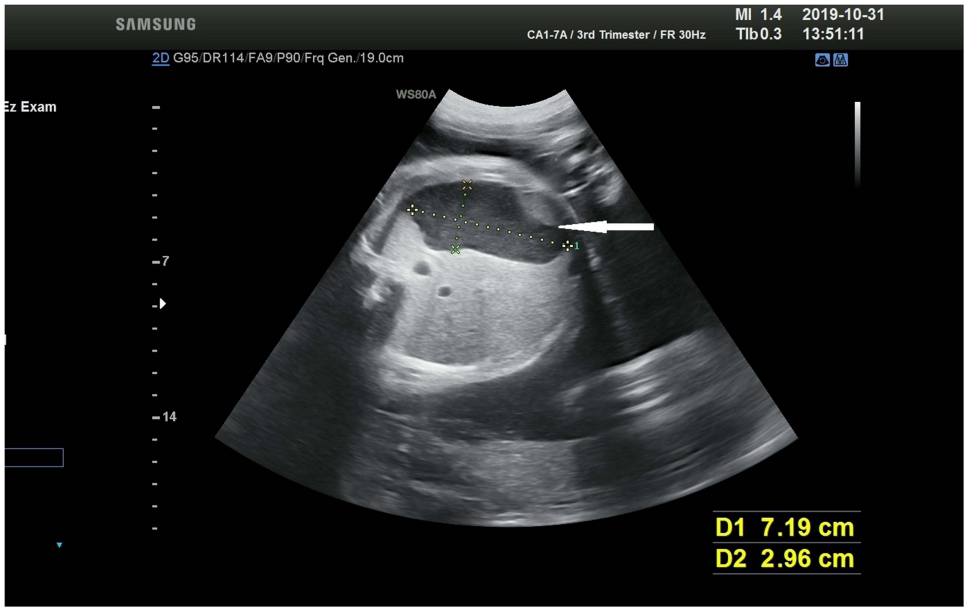

Figure 2 Ultrasonography shows an enlarged fetal stomach bubble.

On physical examination, the neonate was found tachypnea (with respiration rate of 55/min) and absence of skin along Blaschko's lines over the anteromedial aspect of both lower legs almost symmetrically (Figure 3), and also skin absence on the nasal tip (Figure 4), ears, right neck (Figure 5), the right hip, and mucosa absence in the oral cavity, and totally the skin missing covered about 17% of the whole body surface (Roughly, calves, feet, buttocks, and neck accounted for 10%, 3.5%, 2.5%, and 1% respectively), with scattered blisters. The skin and mucous membrane adjacent to these defects appeared "normal" but showed a strongly positive Nikolsky test. Moreover, there was bilateral ear maldevelopment (Figure 5).

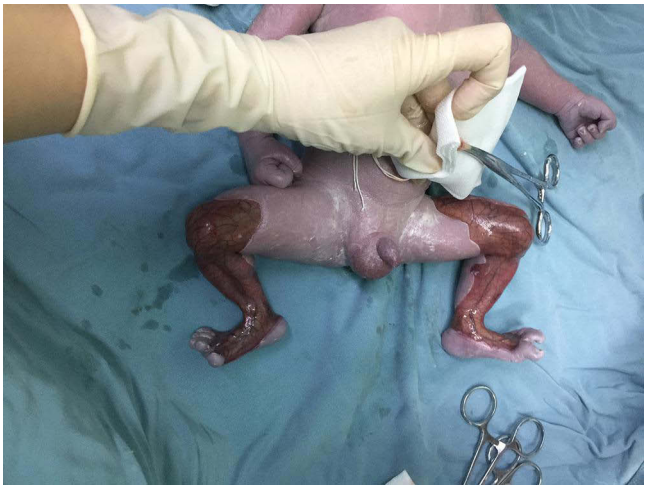

Figure 3 Absence of skin of both lower legs went along Blaschko's lines over the anteromedial aspect starting from the lower third (of right leg) and upper fourth (of left leg) of the thighs and extending to the dorsal and medial plantar aspects of the feet. The lesions had sharply demarcated borders covered by a brownish-red moist ultrathin translucent membrane, beneath which the vasculature was clearly visible.

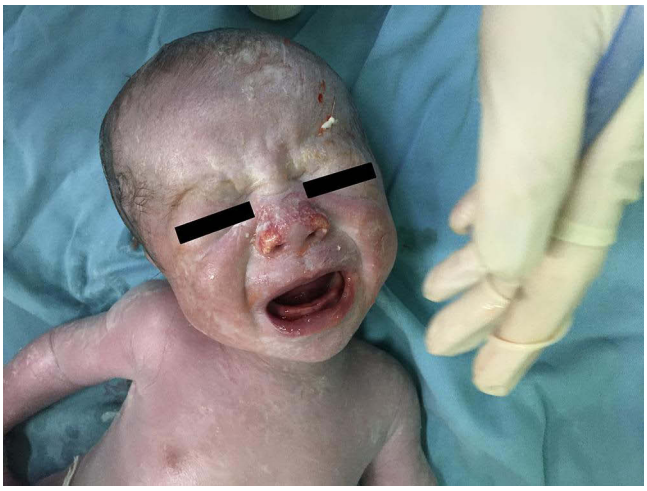

Figure 4 Aplasia cutis congenita(ACC) also exists on the nose.

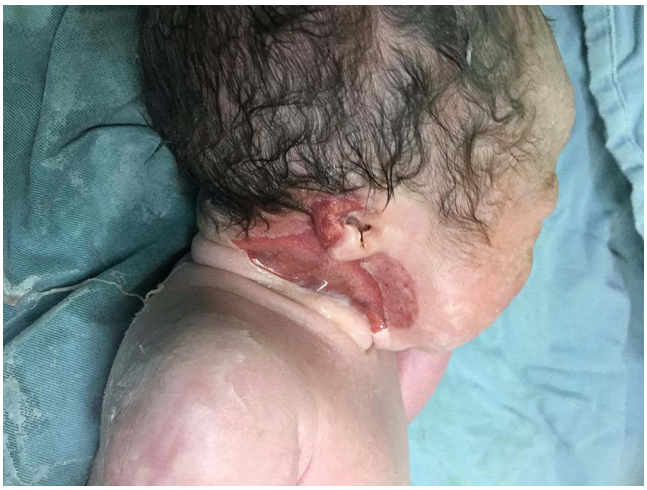

Figure 5 The neonate's external ears were both dysplastic, and aplasia cutis congenita(ACC) also exists on the right neck.

Right after the delivery, routine airway sputum suctioning was performed on the neonate. Subsequently, the neonate was transferred to the Neonatal Intensive Care Unit (NICU) laminar flow ward due to extensive skin lesion and pneumonia, placed in reverse isolation in a 34°C temperature-controlled incubator with humidity of 55%, aiming to minimize insensible water loss. The neonate received immediate thermoregulation and total parenteral nutrition, along with vitamin K1 for vitamin K deficiency bleeding (VKDB) prevention, analgesia, sedation, and cefuroxime anti-infective treatment. In terms of care, continuous electrocardiographic monitoring, oxygen inhalation, and fasting were initiated promptly.

For skin care, given the extensive abnormal skin area of the patient, to alleviate pain and reduce the duration of dressing changes, we planned an alternate-day dressing change strategy, alternating between the trunk and limbs. The dressing sequence involved moistening and softening, wound cleaning, medication application, covering with petrolatum gauze, non-adherent foam dressing, and self-adhesive elastic bandage wrapping.

The neonatologist requested urgent consultations. Given absence of skin and scattered blisters of the skin, the dermatologist suspected "Bart Syndrome". A subsequent plain abdominal radiograph was performed and it revealed a large gastric air bubble with no gas distally (Figure 6), indicating PA. Consequently, the pediatric surgeon made a supplementary diagnosis of CPA. After 26 hours and 53 minutes of hospitalization, the neonate's family decided to discontinue treatment, and the neonate passed away approximately 30 hours after birth. Despite departmental discussions mentioning the possibility of EB and/or ACC in the neonate and proposing the need for pathological and genetic examinations, these planned investigations were not implemented due to the neonate's short lifespan, lack of family support, and other factors.

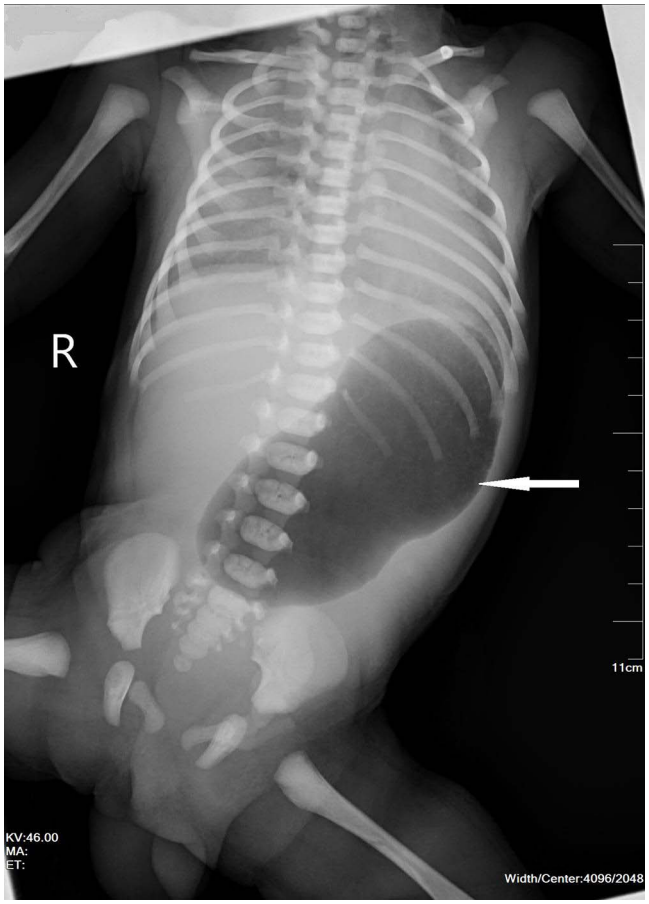

Figure 6 Abdominal X-ray reveals a single gas bubble in a distended stomach with no gas detected in the distal portions, indicating pyloric atresia (PA).

Disscussion

In the management of this rare and critically ill case, we lacked a precise diagnosis and a cutting-edge, therapeutic treatment approach, and the result was poor with the patients demise. However, this scenario is emblematic of the challenges many physicians, hospitals, or even large medical centers face when dealing with rare and severe diseases. We attempt to analyze the reasons for the imprecise diagnosis and poor treatment outcome, and distill lessons from this unsuccessful case, aiming to provide foresight for similar instances in the future.

EB is the prototypic group of disorders with skin fragility defined by blistering from minimal mechanical trauma with disruption at the dermo-epidermal junction with four major subgroups based on the ultrastructural level of blistering in affected tissues: EB simplex, junctional EB(JEB), Dystrophic Epidermolysis Bullosa(DEB), and Kindler syndrome.[9] Overall prevalence and incidence of all major EB types combined in the the National EB Registry (NEBR) were 11.1 per one million population and 19.6 per one million live births, respectively.[10]

PA is characterized by vomiting, failure to tolerate any feeding or to pass stool, and a distended abdomen with a large stomach bubble.[11] It was first described by Scottish surgeon J. Calder in 1749. PA typically occurs in isolation but can also be associated with in 43%-54% of cases. The most common associated anomalies include EB and other bowel atresias, specifically hereditary multiple intestinal atresias (HMIA).[12] And PA can be seen in association with all types of EB, but most commonly with JEB.[13] Otherwise, Carmi Syndrome, occurs in approximately 18% of the cases of PA.[14]

ACC is a rare congenital disorder characterized by a localized absence of skin, dermal appendages, and, in some cases, subcutaneous tissues. It was first described by Cordon in 1767 as a lesion on the extremity and was in 1826 that Campbell reported a case of ACC on the scalp. The reported incidence of ACC varies from one in 10,000 live births to 2.8 in 10,000 newborns, with a predisposition for female patients. However, the incidence is likely much larger, as mild cases of ACC have probably been underreported.[14] Previously, Frieden (1986) classified ACC by location and presence of the other anomalies into nine groups. According to this classification, group 6 was defined as "Bart's syndrome" which is characterized by a combination of congenital localized absence of skin (CLAS).[15] ACC may exist in almost all subgroups of EB.[16]

Bart Syndrome, first described by Bart in 1966, is a rare genetic disorder characterized by ACC and EB.[17] And it may be associated with nail abnormalities such as congenital absence or nail dystrophy,[17,18] which are not absolutely required for making the diagnosis.[15] In 2014, Bart Syndrome was assigned a new proposed name as EB with congenital absence of skin(CAS).[19] Reports have indicated that 30.21% (29/96) of Bart Syndrome patients present with extracutaneous abnormalities, with the most common being PA and ear malformations. Among these cases, 58.62% (17/29) involve PA, and within this subset, 58.82% (10/17) were associated with JEB.[8] Additionally, a systematic review revealed that Bart Syndrome manifested in 28% of patients diagnosed with Carmi Syndrome.[1]

Carmi Syndrome, a rare genetic disorder with PA and EB, had been described in around 100 cases to 2008, primarily reported in the United States (13.7%), India (12.7%), Saudi Arabia (10.8%), Turkey (7.8%), and Japan (6.9%). Nearly half of recorded patients exhibited polyhydramnios on prenatal ultrasound, 76% were preterm infants, and about 25% had a family history of bullous skin disease.[1]

Upon reflection on our management of the patient, we recognize several shortcomings. First and foremost, we mistakenly diagnosed Carmi Syndrome as a combination of Bart Syndrome and CPA. Bart Syndrome and CPA are both rare diseases; therefore, the likelihood of these two rare conditions occurring simultaneously is exceedingly low. Hence, when there is suspicion of their coexistence, we should have considered the possibility of an internal connection within the patient's syndrome, and the syndromes in the patient may represent a singular disease entity. However, neither the dermatologist nor the pediatric surgeon realized that this was a rarer distinct disease entity and, in their respective fields, made an unilateral diagnose. Secondly, both the dermatologist and pediatric surgeon suspected the possibility of "Bart Syndrome" in the newborn, however, the medical records lack information on whether there were deformities in the newborn's nails and toenails. This may be attributed to healthcare personnel either forgetting to document or, alternatively, the physicians simply overlooking examinations of these crucial anatomical sites. Thirdly, in managing the patient, consultations were sought separately from dermatology and pediatric surgery. Unfortunately, there was a lack of communication between the dermatologist and the pediatric surgeon. Precisely, no multidisciplinary team

discussion(MDT) was organized, involving departments of dermatology, pediatric surgery, NICU and so forth. This deficiency contributed to the unilateral nature of our diagnosis. Forthly, Bart Syndrome is mostly an autosomal dominant genetic disorder and lesions are usually unilateral.[15] Nevertheless, in reality, the skin lesions in the patient were essentially symmetrical. And if this diagnosis were accurate, at least one of the newborn's parents should have exhibited related symptoms. However, none of his direct relatives displayed any symptoms associated with Bart Syndrome. As mentioned above, Bart Syndrome is observed in 28% patients with Carmi Syndrome.[1] Therefore, overall, the diagnosis of Bart Syndrome was hasty, unilateral and inaccurate. Last but not least, we did not perform a skin biopsy and genetic testing on the patient, primarily due to the severe condition of the patient, the grave prognosis, and the parents opting to forego treatment and further examinations.

Regarding this case, there are several aspects that require specific clarification. Firstly, non-bilious vomiting was not observed in our case, and we speculate possible reasons as follows: ①. The newborn was kept fasting; ②. During airway suctioning, amniotic fluid from the respiratory tract was aspirated along with gastric contents. ③. The newborn's survival time was too short to manifest this typical symptom. Secondly, in our case, the presence of meconium-stained amniotic fluid, resulting in a yellowish-brown color, suggests the possibility of intrauterine subpyloric defecation behavior of the fetus. We hypothesize that PA may have developed gradually after the secretion of bile. Certainly, the reasons for this phenomenon await further investigation. Thirdly, in this study, the diagnosis of Carmi Syndrome made by the author is straightforward and primarily based on clinical observations, and no specific examinations, such as light microscopy, direct immunofluorescence, TEM, intraoperative dissection, autopsy, etc., were conducted on the patient. Last but equally important, considering the fragile condition of the patient's skin, the optimal site for intravenous catheter placement should have been the umbilical vessel. However, we actually placed the intravenous catheter in the left or right arm for fluid infusion or medication administration, a decision made in the context of neonatal resuscitation. If the newborn's hospitalization duration is prolonged, we would opt for umbilical vessel catheterization to minimize potential damage to the infant's skin.

While the outcome for the patient was unfavorable (which was nearly unavoidable), there were commendable aspects in the diagnosis and treatment of the patient.Firstly, the pregnant woman first visited our obstetrics clinic at 36^{+5} weeks. Before that, local prenatal check-ups were not standardized. She underwent a deferred oral glucose tolerance test (OGTT) around the 32nd week of gestation, a procedure that ideally should have been conducted between the 24th and 28th weeks, with fasting, 1-hour postprandial, and 2-hour postprandial blood glucose levels of 7.13 mmol/L, 7.83 mmol/L, and 5.25 mmol/L, in sequence. However, she was not diagnosed with GDM and did not receive dietary or weight management. When our ultrasound indicated polyhydramnios and an enlarged fetal gastric bubble, the outpatient obstetrician did not automatically attribute it to unmanaged GDM-related polyhydramnios but instead raised suspicions of "fetal digestive tract obstruction." This suspicion served as a crucial alert for subsequent diagnosis and treatment for both the pregnant woman and the newborn. Secondly, due to vigilance regarding digestive tract obstruction, we had a neonatologist present for assistance with neonatal treatment before the newborn's delivery.

In conclusion, from previous literature and our experience with this case, we propose as follows: ①.The possibility of Carmi Syndrome should be ruled out in every neonate with PA regardless of the degree of skin blistering. Similarly, when a patient exhibits clinical manifestations of EB, a complete medical history and physical examination are necessary to determine whether there is extracutaneous involvement and the differential diagnoses must include Bart syndrome and Carmi Syndrome. ②. For rare and/or severe diseases, it is imperative for large hospitals to establish an interdisciplinary clinical committee or multidisciplinary teams(MDTs). In the context of this case, it should have be discussed with all involved disciplines, ie pediatric surgeons, neonatologists and, dermatologists. ③. Carmi Syndrome follows an autosomal recessive inheritance pattern. Given a proband in a family, then genetic counseling and prenatal diagnosis are supposed to be performed prior to subsequent childbearings. ④.In consideration of eugenics and given the high challenge in the treatment of Carmi Syndrome, termination of pregnancy might be contemplated if prenatal diagnosis reveals indicators such as polyhydramnios, gastric enlargement, the "snowflake sign", abnormal external ears, signs of skin desquamation, lower limb anomalies, chorioamniotic membrane separation, and a notable perioral hypoecogenicity identified through ultrasonography.[20,21]

Ethics Statement

The study was approved by the ethics committee of Chengdu Women's and Children's Central hospital (V1.0 2023103).

Declaration of Patient Consent

Written informed consent was obtained from the guardian of the patient for publication of this case report and any accompanying images.

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Disclosure

The authors declared that they had no conflicts of interest to this work.

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