Case Report

Bart Syndrome: A Rare Congenital Disorder with Extensive Aplasia Cutis and Genital Anomalies – A Case Report

Abe George[1], Deepali Chauhan[2], Sumir Kumar[3]

[1]Senior Resident, Department of Dermatology, Guru Gobind Singh Medical College and Hospital, Faridkot, Punjab, India, [2]Junior Resident, Department of Dermatology, Guru Gobind Singh Medical College and Hospital, Faridkot, Punjab, India, [3]Professor and Head, Department of Dermatology, Guru Gobind Singh Medical College and Hospital, Faridkot, Punjab, India

Abstract

Background: Bart syndrome is a rare genetic disorder characterized by a triad of congenital aplasia cutis, epidermolysis bullosa, and nail abnormalities. **Case Report:** We report a full-term newborn born through IVF to a 40-year-old female with multiple bullae, skin detachment, aplasia cutis, nail dystrophy, anonychia, flattened nasal bridge, and external genital anomalies. **Management:** A diagnosis of Bart syndrome was made based on clinical findings and negative blood and pus cultures. The infant was managed conservatively in the neonatal intensive care unit (NICU) with low-flow oxygen, intravenous hydration, temperature regulation, and infection prevention measures. **Conclusion:** The presence of external genital anomalies, bilateral limb involvement, history of IVF, and advanced maternal age in our case of Bart syndrome are uncommon features, highlighting the need for careful evaluation of associated abnormalities.

Keywords: Bart's syndrome; supportive therapy; genital anomalies; advanced maternal age; in vitro fertilization.

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Introduction

Bart syndrome, also known as Aplasia Cutis Congenita type VI with Epidermolysis Bullosa, is a rare genetic disorder that was described for the first time in 1966.It is characterized by congenital absence of skin on the legs, any type of epidermolysis bullosa, nail changes such as congenital nail absence or nail dystrophy, and many other anomalies.[1]

In 1986, Frieden proposed a classification of aplasia cutis congenita, constituting Types I to IX, based on the location of Aplasia cutis and other anomalies [Table 1].[2]

Case Presentation

We report a full-term neonate who was admitted on Day 1 of life in the pediatric emergency department of a tertiary care hospital in North India with fluid-filled lesions and detachment of skin over the body. The baby was born to a 40-year-old female in a non consanguinous marriage through in vitro fertilization (IVF) and LSCS in a private institution. There was no history of any cutaneous disorder in parents and siblings. The pregnancy and delivery were uneventful.

The baby's birth weight was 2400 g, with APGAR scores of 7, 9, and 9 at the first, fifth, and tenth minutes, respectively. The cephalic circumference was 34 cm, chest diameter was 30 cm, heart rate was 140 beats per minute, respiratory rate was 68 breaths per minute, and Capillary Filling Time was < 3 seconds.

On mucocutaneous examination, skin was absent over bilateral lower limbs, periumbilical region, the left forearm, the nose, and right retroauricular region [Image 1]. Erythematous erosions surrounded by peeled off skin over bilateral legs, left foot, forearm, cubital fossa, trunk, and face [Image 2]. Multiple flaccid bullae with a clear to yellowish fluid present over chin and sole of right foot, measuring around 1 x 1 cm [Image 3]. Dystrophy of left hand nails and anonychia over bilateral toe nails (Image 4) and flattening of the nasal bridge present [Image 3]. External genitalia showed more prominent labia minora as compared to labia majora and an absent clitoris [Image 1].

All routine investigations, including blood and pus culture sensitivity, were within normal limits. Histopathology and Direct immunofluorescence could not be performed as the parents refused consent.

Hence, based on the clinical manifestations of aplasia cutis, bullae associated with erosions on the body, nail dystrophy, and a flat nasal bridge, and negative blood and pus cultures, a final diagnosis of Bart Syndrome was made, with characteristic genital anomalies that had not previously been described.

The newborn was managed in NICU in a strictly sterile environment, with adequate intravenous hydration and temperature maintenance. She was put on low-flow oxygen via nasal cannula due to laboured breathing. Regular cleaning of the

Address for correspondence: Dr. Deepali Chauhan, Junior Resident, Department of Dermatology, Guru Gobind Singh Medical College and Hospital, Faridkot, Punjab, India
E-mail: deepalichauhan11@gmail.com

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body with normal saline, followed by a paraffin gauze dressing and topical antibiotic ointment over the erosions, was performed. The neonate survived the treatment and was stable.

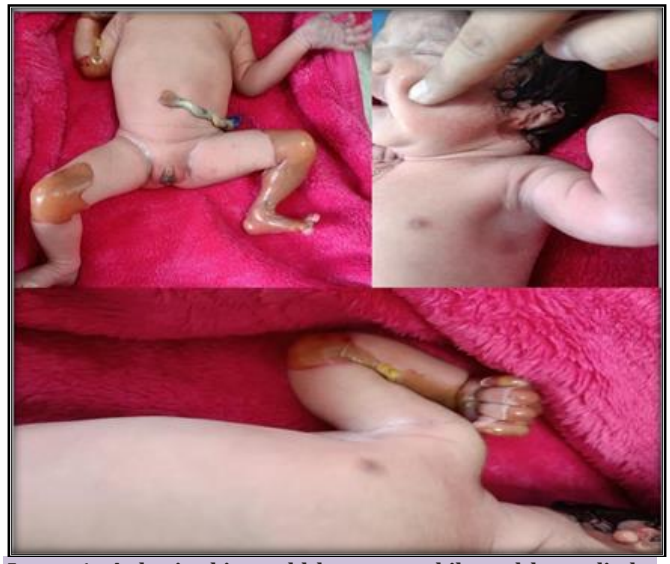

Image 1: Aplastic skin could be seen on bilateral lower limbs extending from distal aspect of right thigh to foot and distal aspect of left thigh to proximal aspect of leg,periumbilical region ,left forearm and behind right ear

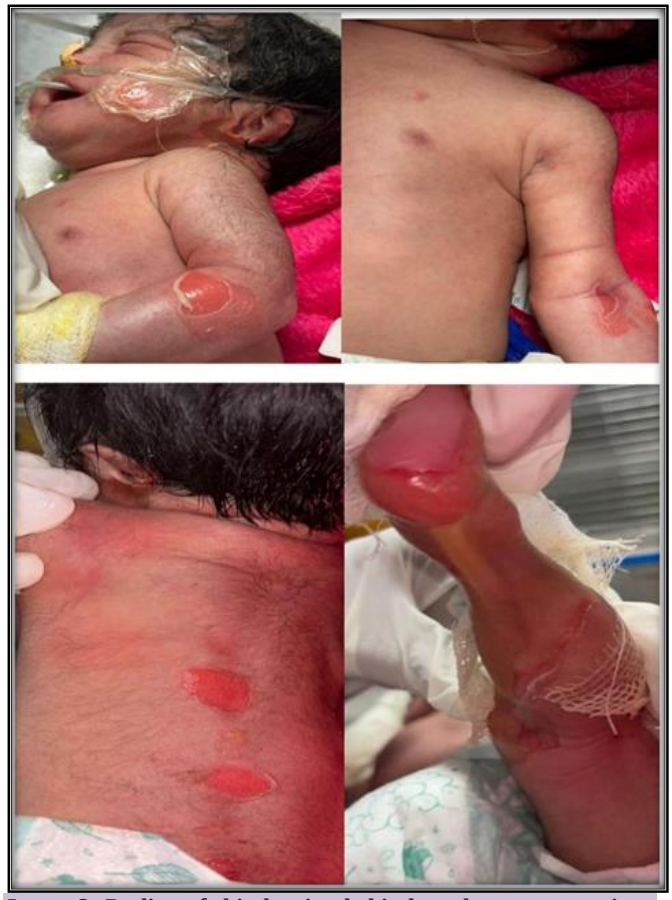

Image 2: Peeling of skin leaving behind erythematous erosions leg, forearm,cubital fossa ,chest,back and cheeks

Image 3: Multiple flaccid bullae with a clear to yellowish fluid present over chin and sole of right foot,measuring around 1 x 1 cm and flattenning of nasal bridge

Image 4: Dystrophy of left hand nails and anonychia over bilateral toe nails

Table 1: Classification of Aplasia

Category	Body area affected	Associated abnormalities	Inheritance
Group 1: scalp ACC without multiple anomalies	Scalp, usually vertex	Cleft lip and palate; tracheoesophageal fistula; double cervix and uterus; patent ductus arteriosus; omphalocele; polycystic kidney	Autosomal dominant or sporadic
Group 2: scalp ACC with associated limb abnormalities	Midline scalp	Limb abnormalities; syndactyly; clubfoot; nail dystrophy; persistent cutis marmorata; encephalocele; woolly hair	Autosomal dominant
Group 3: Scalp ACC with associated epidermal and organoid nevi	Scalp, may be asymmetric	Ocular anomalies; psychomotor retardation	Sporadic
Group 4: ACC overlying embryologic malformations	Abdomen, lumbar skin, scalp; any site	Meningomyeloceles; spinal dysraphia; congenital midline porencephaly; ectopia of ear; omphalocele; gastroschisis	Variable
Group 5: ACC with associated fetus papyraceus or placental infarcts	Multiple, symmetric areas, often stellate or linear, on scalp, chest, flanks, axillae, and extremities	Single umbilical artery; spastic paralysis; nail dystrophy; clubbed hands and feet; amniotic bands	Sporadic
Group 6: ACC associated with epidermolysis bullosa (EB): Blistering, usually localized, without multiple congenital anomalies	Extremities	Blistering of skin and/or mucous membranes; absent or deformed nails; metatarsus varus; congenital absence of kidney	Autosomal dominant or recessive
Group 7: ACC localized to extremities without blistering	Pretibial areas; dorsal aspects of hands and feet	None	Autosomal dominant or recessive
Group 8: ACC caused by specific teratogens	Scalp (with methimazole); any area (with varicella and herpes simplex infections)	Imperforate anus (methimazole); signs of intrauterine infection with varicella and herpes simplex infections	Not inherited
Group 9: ACC associated with malformation syndromes	Scalp; any location	Trisomy 13; 4p– syndrome; Johanson-Blizzard syndrome; focal dermal hypoplasia; amniotic band disruption complex; XY gonadal dysgenesis	Variable

Discussion

Bart syndrome is a rare genetic disorder consisting of aplasia cutis congenita, epidermolysis bullosa, and nail dystrophy.[1] The global incidence of Aplasia cutis congenita is 3 per 10,000 live births.[1]

In most cases of Bart Syndrome, Aplasia Cutis Congenita is unilateral, most often on the lower limbs.[3] However, in our case, it was seen on the bilateral lower limbs, the left forearm, the trunk, and the face. Multiple bullae were seen on the soles of the feet and the chin, along with erosions at trauma-prone sites. Also, dystrophy of nails over the left hand and anonychia over bilateral toe nails were appreciated.

Bart syndrome can be associated with other anomalies, including pyloric atresia, rudimentary ear, flattened nose, wide-set ears, and anomalies of the external genitalia.[1] In our case, the newborn had a broad, flattened nose. External genitalia showed more prominent labia minora as compared to labia majora and an absent clitoris. Very few cases of Bart's syndrome associated with anomalies of external genitalia, like scrotalized and unfused genital bulges with no gonads on palpation, have been reported so far.[1]

ACC is usually sporadic, but autosomal dominant and, less frequently, autosomal recessive cases have been reported.[1] It is associated with the COL7A1 gene on chromosome 3, which codes for collagen type VII.[1] The newborn was born to a 40-year-old female in a non-consanguinous marriage and had no family history of similar symptoms or any other cutaneous disorders in parents and siblings. She was born through IVF and cesarean section in a private institution. In most of the cases reported so far the age of mother is around 30 years.[1] Advanced maternal age in association with Bart's syndrome, as in our case, has not been reported yet. Association of Bart's syndrome with IVF has been reported previously.[4]

Diagnosis of Bart's syndrome is usually clinical, with histopathology and Direct Immunofluorescence studies used to rule out the type of Epidermolysis Bullosa. Also, ultrasound, MRI, karyotyping, and hormonal studies can be done to rule out internal organ abnormalities. In our case, the parents did not give consent for histopathology and genetic testing. So, based on the clinical findings and negative culture reports, a final diagnosis of Bart's syndrome was made.

Overall, the prognosis of Bart's syndrome is good, and the cutaneous lesions recover within 2-3 weeks.[5] However, complications like hypothermia, hypoglycemia, infections, hemorrhage, and fluid and electrolyte imbalance can occur and are the major cause of mortality.[6] Hence, conservative management plays an important role.[1] We managed the newborn in NICU in a strictly sterile environment to avoid the risk of infections, with adequate intravenous hydration and temperature maintenance. She was put on low-flow oxygen via nasal cannula for laboured breathing. Regular cleaning of the body with normal saline, followed by a paraffin gauze dressing and topical antibiotic ointment over the erosions, was performed.

Conclusion

Bart syndrome is a rare genetic disorder characterized by the triad of aplasia cutis congenita, epidermolysis bullosa, and nail dystrophy. Although typically presenting with unilateral lower limb involvement, this case exhibited extensive bilateral involvement along with periumbilical and facial lesions, nail dystrophy, and anonychia. Additionally, the presence of external genital anomalies and a history of IVF and advanced maternal age adds to the uniqueness of this case. We need large-scale

studies to examine the association between IVF and Aplasia cutis congenita. Early diagnosis and meticulous supportive management are crucial to prevent complications such as infections, hypothermia, and fluid imbalance. The unique presentation of this rare syndrome prompted us to report this case.

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Conflicts of interest
There are no conflicts of interest.

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