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Case Report

Aplasia Cutis Congenita: Dilemma of Management in A Resource-Limited Scenario

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Abstract

Aplasia cutis congenital though rare is encountered in clinical practice, presenting varying management dilemma. We present a female neonate delivered at term with congenital absence of skin on both lower limbs, highlighting the constraints of management in a resource-limited environment and the impact on the outcome for the patient.

Keywords: Aplasia Cutis; Management; Outcome; Resource limited

Abbreviations: ACC: Aplasia Cutis Congenita; PCV: Packed Cell Volume; SCBU: Special Care Baby Unit; FBC: Full Blood Count

Introduction

Aplasia Cutis Congenita (ACC), also known as congenital defect of the skin, is a term that describes the absence of skin at birth. It is a relatively rare condition with no more than 125 reported cases as at 1943, 500 cases as at 2013 and often with no other physical abnormalities [1-3]. There is no racial or sex predisposition. Most of the defects manifest as a solitary lesion on the scalp, even though multiple lesions can occur. Though Cordon M gave a description of congenital skin defect involving the extremities, the first account of congenital skin defect was published in 1826 by Willian Campbell (Beresford OD and Samman PD) [4,5]. William Campbell had published the case report of two patients who were siblings, both had scalp skin defect, and both died, one from haemorrhage and the other from hydrocephalus.

Much after that, several cases have been reported. Some of the reported cases were credited to Jones in 1849 of a patient with upper extremity involvement, Priestley in 1859 reported a congenital skin defect over the anterior fontanel about the size of a shilling (Abt IA), Terruhn in 1930 whose review showed that the scalp was involved in 79 cases and other sites in 29 cases, Freud et al in 1945, Beresford OD and Samman PD of three unusual cases, and in recent times by Maillet-Declerck M, et al., who reviewed 29 cases, and a few other case reports [5-7]. The aplasia typically involves only the epidermis, but may also involve the dermis, subcutaneous tissue, bone in rarer cases, and all the way down to the dura on the scalp [8]. Multiple classification systems have been devised; however, the most widely adopted system is the Frieden Classification System [9]. We present a case of aplasia cutis congenita type 6 with epidermolysis bullosa that we manage in our facility, with the aim of reminding the readers that even though it is a rare condition, it does exist, while also highlighting the challenges we encountered in the patient's management.

Case Report

Female neonate delivered at term (37 weeks and 5 days) via an emergency lower segment caesarean section on account of foetal distress to a 29-year old primigravida who resides in South-West Nigeria. Birth weight was 2.55kg with APGAR score of 8 and 9 in the first and fifth minutes respectively. The antenatal period was uneventful. Pregnancy was booked in the first trimester. No history of drug use or fever or any illness during pregnancy reported. Mother was a healthy person with no history of hypertension, diabetes, asthma or sickle cell disease. Family history was negative for similar conditions. Examination revealed a neonate lying calmly in a bed, afebrile, anicteric, well hydrated.

Her vitals were stable with heart rate of 162 beats per minutes and respiratory rate of 52 cycles per minute. On examination of the chest, the patient had vesicular breath sounds; no cardiac murmurs were noted. She had a well-demarcated area of full-thickness skin loss on the anterior surface of the knees extending to the proximal third of the dorsum of the feet bilaterally, with the involvement of the lateral aspect of the plantar surface. The lesions on the leg were almost circumferential. (Figures 1 and 2) show the lesions on the lower limbs at birth and on the 5th day of birth respectively.

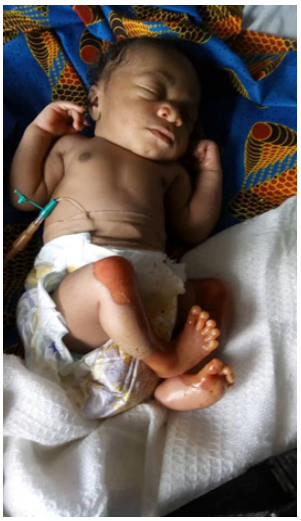

Figure 1: The neonate at birth showing the skin defects over the right knee and the involvement of both feet. There was no active bleeding at the skin defects.

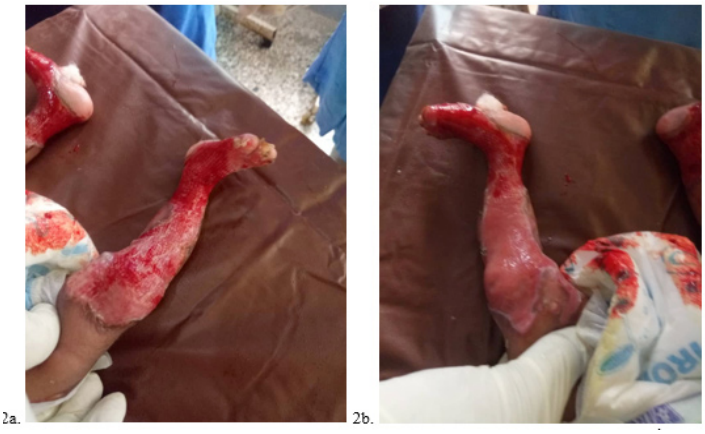

Figure 2: The skin defects on right and left lower limbs respectively on the 5th day of birth. The defects bled actively on change of dressing.

There was no active bleeding from the skin defects at birth. Her anterior and posterior fontanelles were patent and normotensive. The patient had complete Monro reflex, good grasp, rooting and suck reflexes. She was admitted into the Special Care Baby Unit (SCBU) and the plastic surgical team was invited for co-management. Initial investigations at birth that included a Complete Blood Count (CBC), serum electrolytes, Urea and Creatinine (E/U/Cr) were within normal limits. She was started on alternate day dressing with saline and framycetin sulfate petroleum impregnated gauze (sufratulle) dressing; however, it was noted that she was bleeding excessively during dressing change that necessitated frequency of dressing to be reduced to once a week. Repeat CBC was remarkable for thrombocytopenia (platelet count 43 x 10^9 cells per L). She had also developed blisters in the skin around areas where blood samples were collected, as well as the dorsum of the left hand with intravenous access, the buccal mucosa, and patchy areas on the trunk, with the nails exfoliating.

The patient had several blood transfusions for severe anaemia. She was planned for auto-graft / maternal allograft split-thickness skin graft. However, at the age of 31 days, she was discharged home to the out-patients' clinic since her parents were not able to afford hospital fees. Twenty days after discharge during the outpatient visit, the baby was readmitted for anaemia, with a Packed Cell Volume (PCV) of 24%. She was lethargic, afebrile with a heart rate of 160 beats per minute and respiratory rate of 78 per minutes. On examination, it was noted that wound was approximately 70% re-epithelialized. She had a total of 75mls of blood transfused and discharged the following day. One week later at the plastic surgery outpatient clinic follow up, the patient had PCV of 42%. Unfortunately, she was brought to the children emergency room 10 days after last follow up with no cardiac and respiratory activities and pronounced dead at the age of 61 days.

Discussion

Aplasia cutis congenita is a rare condition which mostly involves the scalp in about 70 - 80% of cases, where it can extend to the galea, the pericranium, the bone, and the dura mater [7,10]. Other sites can, however, be involved, such as the forearms, knees, both sides of the trunk, and neck, in decreasing order of frequency [11]. When non-scalp locations are involved, they are often bilaterally symmetrical [12]. Our patient presented with skin defects on upper and lower limbs that were bilaterally symmetrical. There were associated bullae and blistering in adjacent / junctional area which represent a bullous variant of the ACC. The aetiology of ACC is uncertain, but in about one-third of cases, congenital skin defects may be associated with other congenital malformation like trisomy 13, cleft lip and palate, hands and feet defects.

Tempark and Shwayder pointed out contributory factors to aplasia to include genetics as may be seen in association with organoid or epidermal naevi and familial cases with autosomal dominant or recessive inheritance; non-syndromic cases associated with a heterozygous mutation in the BMS1 gene (611448) on chromosome 10q11; placental infection during pregnancy such as varicella (chickenpox) or herpes simplex; ingestion of teratogens

such as methimazole, carbimazole, misoprostol, diclofenac sodium, cocaine, marijuana, and valproic acid; arrest of skin development in the embryo/foetus; amniotic bands; and intrauterine death of a twin foetus [13,10]. Greig had advanced that arrest of development was mostly responsible and tried to justify the belief by noting in favour the usual mid-line position on the scalp, the striking symmetry in other areas, and the association with other congenital defects [5].

We could not pinpoint a cause or risk factor in our patient. There was no history of consumption of any of the teratogens above, neither was there any significant infection during pregnancy. There were no associated anomalies on clinical examination. Small defects typically heal spontaneously over time. In some cases, it has been demonstrated that healing has started in utero. However, larger skin defects present a management dilemma. There is no consensus as to how to manage such large skin defects because of associated risks, but both conservative and surgical approaches had been offered in the early management of the skin defect [12]. Such surgical approaches include skin grafting, though Maillet-Declerck M, et al. had proposed a therapeutic strategy based on the size of the skin defect and the nature of underlying exposed structures [7].

In our case, the patient had been prepared to have conservative care followed by surgical intervention of a combination of meshed auto-graft and sheet allograft (from mother), however, that was not achieved due to unsurmountable management dilemma. The parent could not afford the fee for surgery and continued hospital admission. This is coupled with the inability to afford the money for the instrumentation required for the surgery. The parents were not on any insurance scheme, as such all payments had been out-of-pocket. High out-of-pocket expenditure by citizens as well as inequitable and unsustainable health care financing had been noted to be great challenges to health care [14]. The patient had to be discharged home on dressing and to be followed up in the clinic. Skin grafting had been shown to be of immense value in the management of large skin defects of aplasia cutis congenita. Trah, et al. described a patient with an extensive area of ACC who was treated by Integra®-Dermal Regeneration Template and Split-Thickness Skin Grafting (STSG) [15]. They postulated that the treatment option could prevent severe infection, compartment-syndrome-like conditions, and deformities, as well as prevent new blistering.

Unfortunately, their patient died only 4 days after STSG, and as such long-term benefit could not be proven. Lonie, Phua and Burge reported the management of a large area of ACC on newborn's scalp with a single skin allograft application and noted that it led to complete epithelialization of the skin defect. Liu Y, et al. reported the usage of large - sized thin split - thickness skin grafting in 16 patients with lesion >10% of the Total Body Surface Area (TBSA) [16]. They observed early wound closure and reduced long term scarring. In our case, the planned skin grafting could not be done, but we were equally mindful of the challenges that may be associated with a surgical approach. Beyond the financial challenge noted above, these challenges include risk of possible additional multiple blood transfusion, infection, anaesthetic complications occasioned by general anaesthesia with endotracheal intubation, and prolonged hospital stays.

Having chosen a conservative treatment in view of the resource constraint setting, the other dilemma was that of the choice of dressing material. Several materials have been utilized to aid healing in ACC with satisfactory results. Studies have demonstrated the use of basic fibroblast growth factor application for large ACC of the scalp with bone defect, acellular dermal matrix for ACC on the scalp, a water-vapour permeable polyurethane film (Omiderm®) used alongside with absorbent fine mesh gauze impregnated with 3% bismuth tribromophenate for cranial defect (Canter HI, Vargel I, Nasir S, Kayikcioglu A), ionised silver-coated dressings such as Acticoat® and Aquacel® Ag, as well as antibiotics impregnated petroleum-based gauze [17-20].

We had settled for the latter in this case with the use of sulfratulle, as the patients could not afford more expensive dressing materials. Even though the patient's wound was doing well with the dressing, she had to be transfused repeated due to anaemia from bleeding at dressing times. Dressing change interval was increased to once weekly to mitigate against the loss of blood. Other dressing materials such as the ionised silver-coated dressing would have made less frequent dressing change possible, but the patient could not afford it. Invariably, the patient had to be discharged for out-patient management as the parent could no longer afford the in-hospital care. She, however, died from severe anaemia despite the marked improvement in the wound condition. Chances are that the patient may have survived and received more timely resuscitation if not for the parents' insistence on out-patient care.

Conclusion

While surgical management with split skin graft would have been the best choice of treatment of large multiple ACC as in this patient, the challenges associated with care in a resource-constrained environment may necessitate a reliance only on conservative management with healing by secondary intention. Close infection control by strict adherence to aseptic procedure and prophylactic antibiotics, as well as timely optimization of the packed cell volume will be of utmost importance in achieving a favourable result.

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