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Aplasia Cutis Congenita. A Case Report and Review of Literature

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Aplasia cutis congenita is a birth defect characterised by absence of skin and, in some cases, the subcutaneous tissues. This is a case report of the initial conservative management of a neonate in a rural hospital who presented with aplasia cutis congenita involving the trunk and the lower limbs. The literature is reviewed and the case discussed.

Key words: Aplasia cutis congenital, rural, hospital

Introduction

Aplasia cutis congenita (ACC) is a rare congenital defect characterised by localized absence of skin and in some cases the subcutaneous tissues. It was first described by Cordon in 1767 and later classified by Frieden based on the number and site of the lesions as well as the presence or absence of associated malformations[1]. The most common site of involvement is the scalp but the lesions may occur in any part of the body including the trunk and the extremities[2]. In most cases, aplasia cutis congenita occurs as a solitary defect but it may be associated with other congenital abnormalities[3].

The condition is congenital and therefore presents at birth or in utero. Racial predilection has not been described nor has any sex predilection been found except in cases associated with X-linked malformations. Most cases are sporadic but familial occurrences have been documented[4]. The aetiology is largely unknown and is thought to be multi-factorial[3]. Management is multidisciplinary. The mainstay of treatment is conservative whereby the lesions usually heal by scarring. Surgical intervention also has a role particularly after failed conservative management as well as for delayed reconstructive and cosmetic purposes.

A large amount of literature is available on the topic but it mostly describes the management of this condition in resource-rich set ups. Additionally, most of the literature discusses scalp lesions which are the most common. This is a case report of a neonate managed for ACC involving the trunk and lower limbs in a rural hospital.

Case Report

A neonate noted to have truncal and lower limb skin defects was received in the hospital New Born Unit directly from theatre after being born via emergency caesarean section. The indication for caesarean section was 3 previous scars in labour and non-reassuring foetal status as evidenced by meconium grade 3 stained liquor on vaginal examination.

The birth weight was 2.4 kg and Apgar Scores were 9 at 1, 10 at 5 and 10 at 10. Gestational age was unknown as the mother was unsure of her dates.

Antenatal Clinic (ANC) History: Mother attended antenatal clinic 5 times and the ANC profile done was essentially normal – haemoglobin was 11.4g/dl, VDRL was non reactive, P24 MARKERS were non reactive, mother's blood group was AB+ and urinalysis done was normal. The mother received Tetanus Toxoid 5 during the pregnancy. There was no history of maternal illness, maternal drug use or premature rupture of membranes. She however reported a similar lesion in the upper limb of her third born which healed spontaneously. No documentation or further history was available.

The truncal skin defect was symmetrical extending from just below the nipple superiorly to below the umbilicus inferiorly. There was an island of normal skin around the umbilicus with an associated umbilical hernia. Blood vessels and underlying structures were visible through a translucent membrane. Similar bilateral lesions involving the thighs symmetrically were also noted.

The neonate was otherwise found to be healthy. The child was kept well hydrated and warm in the New Born Unit where the wounds were cleaned and dressed daily with Silver Sulfadiazine and Sufratulle. Infection control procedures were practised with use of intravenous antibiotics and blood transfusion was done with packed cells as necessary. Physiotherapy was done daily and the healing process was reviewed frequently by the surgical team. She is currently in good general condition and doing well in the New Born Unit. The lesions are healing well and the child is gaining weight.

Figure 1a. Neonate with Aplasia Cutis Congenita Type V

Figure 1b. Neonate with Aplasia Cutis Congenita Type V

Discussion

Aplasia cutis congenital is a rare congenital disorder of the skin. Frieden classified it into 9 groups (see TABLE 1) according to the number, location and pattern of lesions and presence or absence of associated abnormalities and mode of inheritance[1]. Type V is ACC involving the trunk and extremities associated with foetal papyraceous or placental infarct. Most of the literature describes Type V as having extensive truncal and limb aplasia with foetal papyraceous or placental infarct[5 6]. Although this case does not have foetal papyraceous or placental infarct it fits best in Type V. Although less common, similar cases are documented in literature and were likewise classified as Type V[7 8].

In this case no obvious aetiopathogenesis could be described. Proposed etiology includes genetic predisposition, intrauterine vascular ischaemia, amniotic bands, drugs such as methimazole, cocaine and benzodiazepines, maternal viral infection and nutritional and vitamin deficiencies[9-13]. Although the majority of cases are sporadic, familial occurrences have been described[4 14] and familial history of a similar lesion in another sibling suggests a possible genetic factor.

This neonate was managed conservatively with the basic care of a new born taking priority. This can be instituted in any basic hospital set-up although we chose to manage ours in the controlled environment of the new born unit with the aim of infection control as well as specialized care from the neonatal nurses. Both conservative and operative treatment have a role in the management of

ACC. Conservative management includes cleaning and dressing with Silver Sulfadiazine and other antiseptic ointments, saline dressings and continuous saline drips.

Table 1. Classification Scheme for Aplasia Cutis Congenita[1]

Group 1	**Scalp lesions without multiple anomalies**
Group 2	**Scalp lesions with associated limb abnormalities**
Group 3	**Scalp lesions associated epidermal and sebaceous naevi**
Group 4	**Aplasia cutis congenita overlying deeper embryologic malformations such as meningomyelocele, cranial stenosis, spinal dysraphism, gastroschisis, and omphalocele.**
Group 5	**Truncal and limb lesions with associated foetus papyraceus or placental infarcts**
Group 6	**Aplasia cutis congenita associated with epidermolysis bullosa**
Group 7	**Aplasia cutis congenita localized to the extremities without epidermolysis bullosa**
Group 8	**Aplasia cutis congenita caused by teratogens**
Group 9	**Aplasia cutis congenita with associated malformation syndromes**

The goal of conservative management is to allow granulation and healing by secondary intention as well as to avoid potential operative risks[15]. Operative treatment options are mainly indicated for ACC involving the scalp and they include primary closure, skin grafting, flaps with or without tissue expansion and cranial vault reconstruction using split rib grafts[15].

The mainstay of management of type V aplasia cutis congenita is conservative[16 18] with daily cleaning and dressing of wounds and use of antibiotics in the case of infection. Lesions are expected to heal spontaneously by contracture. There is good response to conservative treatment even in cases with involvement of large areas of the body surface[19]. Surgical intervention may play a role at a later date particularly for scar correction. There is little role for early surgical intervention. This neonate was managed with daily cleaning and dressing of the skin lesions with both silver sulfadiazine and sufratulle. Infection control practices were instituted and a course of intravenous antibiotics administered when the child showed signs of infection.

The management was multidisciplinary with early physiotherapy and frequent surgical reviews of the healing process.

Conclusion

Aplasia cutis congenital is a rare congenital skin defect that can easily be managed in a limited resource setting. The management is conservative with daily cleaning and dressing of the lesions. Surgical intervention may be required and in those cases is mostly reconstructive and/or cosmetic.

References

1. Frieden IJ: Aplasia cutis congenita: a clinical review and proposal for classification. *J Am Acad Dermatol*, Apr 1986; 14(4):646-60
2. Morelli JG: Cutaneous Defects. In *Nelson Textbook of Pediatrics*. 18th edition. Philadelphia, PA: Saunders Elsevier; 2007:647
3. Buchel T, Devaul W, Frey K: Pop Quiz: Newborn with Scalp Lesion. *American Family Physician*. October 15, 2005. Available at: http://www.aafp.org/afp/2005/1015/.
4. Lestringant G, Al Towairky A: (1989): Three Siblings With Extensive Aplasia Cutis Congenita of the Scalp and Underlying Bone Defect: Autosomal Recessive Inheritance. *International Journal of Dermatology*, 28: 278–279. doi: 10.1111/j.1365-4362.1989.tb04830.x
5. Klein RQ, Robinson DM, Lieber CD, Antaya RJ: Symmetric Aplasia Cutis Congenita Associated with Fetus Papyraceus: Report of Two Cases. Article first published online: 25 mar 2011. doi: 10.1111/j.1525-1470.2011.01314.x
6. Wadams S, Garrett-Cox R, Kitteringham L: Aplasia cutis in association with a triplet pregnancy and fetus papyraceus
7. Cambiaghi S, Schiera A, Tasin L, Gelmetti C: Aplasia Cutis Congenita in Surviving Co-twins: Four Unrelated Cases. *Pediatric Dermatology*.Vol. 18, No. 6:511±515, 2001
8. Boente MC, Frontini MV, Acosta MI, Saleme C, Barrionuevo S, Asial R: Extensive Symmetric Truncal Aplasia Cutis Congenita without Fetus Papyraceus or Macroscopic Evidence of Placental Abnormalities. *Pediatric Dermatology*. Vol. 12 No. 3 228-230
9. Izhar R, Ghani T: Aplasia cutis congenita and antithyroid drugs. *J Pak Med Assoc*. Nov 2002; 52(11):526-8.
10. Karg E, Bereg E, Gaspar L, Katona M, Turi S: Aplasia cutis congenita after methimazole exposure in utero. *Pediatr Dermatol*. Jul-Aug 2004; 21(4):491-4.
11. Zhou J, Zheng L, Tao W: Systemic aplasia cutis congenita: A case report and review of the literature. *Pathology – Research and Practice* 206 (2010) 504–507
12. Rasheed T, Orlando A, Gordon DJ: Basal cell carcinoma in aplasia cutis congenita. *British Journal of Plastic Surgery* (2000), 53, 2000 The British Association of Plastic Surgeons doi: 10.10541bjps.2000.3337
13. Jonathan Kantor, Albert C. Yan, Chad M. Hivnor, Paul J. Honig, Richard Kirschner et al : Extensive Aplasia Cutis Congenita and the Risk of Sagittal Sinus Thrombosis *(Reprinted) Arch Dermatol/Vol 141, May 2005*
14. Casanova D, Amar E, Bardot J, Magalon G: Aplasia cutis congenita: Report on 5 family cases involving the scalp. *Eur J Pediatr Surg*. Aug 2001; 11(4):280-4.
15. Theile RJW, Lanigan MW, McDermant GR: Reconstruction of aplasia cutis congenita of the scalp by split rib cranioplasty and a free latissimus dorsi muscle flap in a nine month old infant. *Brlrish Journal of Plastic Surgery* (l995),48, 507-510 Q 1995 The British Association of Plastic Surgeons
16. Bigliardi PL, Braschler C, Kuhn P, Sigrist J, Buechner S, Rufli T: Unilateral aplasia cutis congenita on the leg.

17. Ozek C, Gurler T, Alper M, Gorken C, Songur E: Aplasia cutis congenita with epidermolysis bullosa: a case report.
18. Bang RL, Ghoneim IE, Gang RK, Al Najjadah: Treatment dilemma: conservative versus surgery in cutis aplasia congenita. *Eur J Pediatr Surg.* 2003; 13(2):125-9 (ISSN: 0939-7248)
19. Suliman MT, Quazi A: Case Report: Aplasia cutis congenita of the trunk in a Saudi newborn. The British Association of Plastic Surgeons (2004) 57, 582–584