

A Rare Case of Aplasia Cutis Congenita

Mostafa Dahmardehei

Plastic Surgeon, Ali-Ebn-Abitaleb Hospital, Zahedan University of Medical Sciences, Zahedan, Iran

Received: 15 May 2012; Received in revised form: 10 Dec. 2012; Accepted: 5 Jan. 2013

Abstract- Aplasia cutis congenita is a rare anomaly presenting with the absence of skin. No definite etiology is available. The most common site is the scalp. We present an instance with ACC occurring symmetrically in both sides of the body from chest to flank.

© 2013 Tehran University of Medical Sciences. All rights reserved.

Acta Medica Iranica, 2013; 51(5): 341-344.

Keywords: Aplasia cutis congenital; Giant; Nonscalp

Introduction

Aplasia cutis congenita (ACC) is an uncommon anomaly of absence of skin. It may be localized or widespread. It most commonly presents as a solitary lesion of the scalp and may be as large as 70 percent of that site.

Although usually benign, they may be associated with other physical abnormalities and syndromes. Frieden classified them into 9 groups based on the number and the presence or absence of other anomalies (1). Nearly 86 percent belong to the first group with a solitary lesion. Involved areas are well-circumscribed, not inflamed, and vary in size from 0.5 to 10 cm or larger. At birth lesions may appear as scars or ulcers (1). On the scalp they may appear as parchment-like scars with alopecia.

The cause is not certain as it is the result of more than one disease process. Genetic factors, teratogens, compromised vasculature to the skin, and trauma are all implicated (1,2). There is no racial or sexual predilection. It is present from birth.

Maximum tensile force during the development of scalp hair whorl is implicated for the scalp lesion. Early rupture of amniotic membrane forming amniotic bands may also be responsible (1).

According to our knowledge, there is not any report of symmetric body involvement around the world. We present an instance with ACC occurring symmetrically in both side of the body from chest to flank.



Figure 1. Anterior View of the patient.

Case Report

Clinical synopsis

A 3-day-old girl of nonconsanguinous parents presented at Ali-ebn-abitaleb Hospital (Zahedan, Iran) for a skin lesion that was present on both sides of her body birth. The lesions were quadrangular ulcers extending from the chest anteriorly to the flank posteriorly. Examination revealed to 15 x 5 cm ulcers within which there was a total absence of skin (Figures 1, 2).

There were no other organ abnormalities on clinical examination. Radiological examination and ultrasonography of the abdomen revealed no abnormalities. Routine investigations were within normal limits.

Corresponding Author: Mostafa Dahmardehei

Plastic Surgeon, Ali-ebn-abitaleb Hospital, Zahedan University of Medical Sciences, Zahedan, Iran
Tel: +98 541 3424392, Fax: +98 541 3424391, E-mail: dahmardehei@zaums.ac.ir



Figure 2. Posterior view of the patient.

Discussion

Aplasia cutis congenita (ACC) is an uncommon anomaly of absence of skin. Frieden (1) created a classification system for aplasia cutis congenita consisting of 9 groups based on the number and location of the lesions and the presence or absence of associated malformations:

Group 1-This is scalp aplasia cutis congenita without multiple anomalies (3). Nearly 86% of all solitary lesions occur on the scalp. A collar of hair is often seen around the defect. It can be autosomal dominant (4) or sporadic (5).

Group 2-This is scalp involvement with limb anomalies.(6, 7, 8, 9) Adams-Oliver syndrome(10-14) is a distinct subtype in which distal limb reduction abnormalities are found in association with solitary midline scalp defects. More than 15 such cases have been reported, usually with an autosomal dominant inheritance pattern and variable genetic expression. The scalp lesions tend to be large. The most common limb malformation is hypoplastic or absent distal phalanges. Other anomalies may include cutis marmorata telangiectatica congenita, hemangiomas, cranial arteriovenous malformation, skin tags, supernumerary nipples, and woolly hair.

Group 3-This is scalp aplasia cutis congenita with epidermal and sebaceous (organoid) nevi (15,16), which also involve the scalp, usually adjacent to the cutis aplasia. Some patients have also had ophthalmic and neurologic findings typical of epidermal nevus syndrome, including seizures, mental retardation, corneal opacities, and eyelids colobomas. Inheritance is sporadic.

Group 4-This is aplasia cutis congenita often with a

hair collar overlying deeper embryologic malformations.(17-20) Examples include meningomyelocele, pencephaly, leptomeningeal angiomas, cranial stenosis, spinal dysraphism, gastroschisis, and omphalocele. The inheritance pattern in this group varies with the associated underlying condition.

Group 5-This is aplasia cutis congenita associated with fetus papyraceus or placental infarct (21-25). Extensive truncal and limb aplasia cutis congenita in a linear or stellate configuration is associated with the presence of fetus papyraceus. Fetus papyraceus is found at the time of delivery and results from the death of a twin fetus early in the second trimester. The surviving fetus is affected with aplasia cutis congenita and usually is otherwise normal.

Group 6-This is aplasia cutis congenita associated with simplex, junctional, or dystrophic types of epidermolysis bullosa (EB) (26-28). Many reports describe aplasia cutis congenita, usually occurring on the lower extremities, in patients eventually diagnosed with EB. Initially described as Bart syndrome, this type of presentation represents a variant of dystrophic EB. A subgroup includes the association of pyloric or duodenal atresia, ureteral stenosis, renal abnormalities, craniofacial abnormalities, nail dystrophy, and aplasia cutis congenita.

Group 7-This is aplasia cutis congenita localized to the extremities without EB (29-31). At least 2 families have been reported in which multiple members have had extensive aplasia cutis congenita on the pretibial lower extremities and the dorsal aspects of the hands and feet.

Group 8-This is aplasia cutis congenita due to teratogens. A few cases of aplasia cutis congenita have been linked to intrauterine infection with herpes simplex virus or varicella-zoster virus or to exposure to methimazole (32-35) in the treatment of maternal thyrotoxicosis during pregnancy. Imperforate anus has been associated with methimazole or carbimazole exposure during gestation.

Group 9-This is aplasia cutis congenita associated with malformation syndromes (36,37). Aplasia cutis congenita has been reported as a characteristic in many syndromes and more will be reported. Various syndromes (38-41) and dysplasias include trisomy 13 (Patau syndrome) with large membranous scalp defects, 4p- (Wolf-Hirschhorn) syndrome with midline scalp defects, Setleis syndrome with bitemporal aplasia cutis congenita and abnormal eyelashes, Johanson-Blizzard syndrome with stellate scalp defects, focal dermal hypoplasia (Goltz syndrome), amniotic band disruption

complex, oculocerebrocutaneous (Delleman) syndrome, scalp-ear-nipple syndrome (Finlay-Mark syndrome), Kabuki syndrome (42), and 46XY gonadal dysgenesis. Reticulolinear aplasia cutis congenita on the face and the neck is a distinctive cutaneous manifestation in several syndromes linked to Xp22.

As mentioned above, there is no report of symmetric body involvement around the world. We suggest adding another group to Frieden's classification system to include this rare form of ACC.

References

- Frieden IJ. Aplasia cutis congenita: a clinical review and proposal for classification. *J Am Acad Dermatol* 1986;14(4):646-60.
- Kruk Jeromin J, Jainik J, Rykala T. Aplasia Cutis Congenita of the scalp: Report of 16 cases. *Dermatol Surg* 1998; 24 (5):549-53.
- Caksen H, Kurtoglu S. Our experience with aplasia cutis congenita. *J Dermatol*. Jun 2002;29(6):376-9. Moros Pena M, Labay Matias M, Valle Sanchez F. (Aplasia cutis congenita in a newborn: etiopathogenic review and diagnostic approach). *An Esp Pediatr* 2000;52(5):453-6.
- Drolet B, Prendiville J, Golden J, Enjolras O, Esterly NB. 'Membranous aplasia cutis' with hair collars. Congenital absence of skin or neuroectodermal defect?. *Arch Dermatol* 1995;131(12):1427-31.
- Colon-Fontanez F, Fallon Friedlander S, Newbury R, Eichenfield LF. Bullous aplasia cutis congenita. *J Am Acad Dermatol*. May 2003;48(5 Suppl):S95-8.
- Benjamin LT, Trowers AB, Schachner LA. Giant aplasia cutis congenita without associated anomalies. *Pediatr Dermatol* 2004;21(2):150-3.
- Chitnis MR, Carachi R, Galea P. Familial aplasia cutis congenita. *Eur J Pediatr Surg* 1996;6(2):100-1.
- Fagan LL, Harris PA, Coran AG, Cywes R. Sporadic aplasia cutis congenita. *Pediatr Surg Int* 2002;18(5-6):545-7.
- Davidson AW, Hosalkar HS, Hill RA, Monsell F. Radial dysplasia with localized cutis aplasia congenita. *J Pediatr Orthop B* 2003;12(6):398-401.
- Balasubramanian M, Collins AL. Aplasia cutis congenita, terminal limb defects and periventricular leukomalacia in one sibling with minor findings in the other-probable autosomal recessive Adams-Oliver Syndrome. *Eur J Med Genet* 2009;52(4):234-8.
- Bilginer B, Onal MB, Bahadir S, Akalan N. Aplasia cutis congenita of the scalp, skull and dura associated with Adams-Oliver syndrome. *Turk Neurosurg* 2008;18(2):191-3.
- Dyall-Smith D, Ramsden A, Laurie S. Adams-Oliver syndrome: aplasia cutis congenita, terminal transverse limb defects and cutis marmorata telangiectatica congenita. *Australas J Dermatol* 1994;35(1):19-22.
- Orstavik KH, Stromme P, Spetalen S. Aplasia cutis congenita associated with limb, eye, and brain anomalies in sibs: a variant of the Adams-Oliver syndrome? *Am J Med Genet* 1995;59(1):92-5.
- Temtamy SA, Aglan MS, Ashour AM, Zaki MS. Adams-Oliver syndrome: further evidence of an autosomal recessive variant. *Clin Dysmorphol* 2007;16(3):141-9.
- Lam J, Dohil MA, Eichenfield LF, Cunningham BB. SCALP syndrome: sebaceous nevus syndrome, CNS malformations, aplasia cutis congenita, limbal dermoid, and pigmented nevus (giant congenital melanocytic nevus) with neurocutaneous melanosis: a distinct syndromic entity. *J Am Acad Dermatol* 2008;58(5):884-8.
- Neri I, Savoia F, Giacomini F, Raone B, Aprile S, Patrizi A. Usefulness of dermatoscopy for the early diagnosis of sebaceous naevus and differentiation from aplasia cutis congenita. *Clin Exp Dermatol* 2009;34(5):e50-2.
- Kantor J, Yan AC, Hivnor CM, Honig PJ, Kirschner R. Extensive aplasia cutis congenita and the risk of sagittal sinus thrombosis. *Arch Dermatol* 2005;141(5):554-6.
- Kim CS, Tatum SA, Rodziewicz G. Scalp aplasia cutis congenita presenting with sagittal sinus hemorrhage. *Arch Otolaryngol Head Neck Surg* 2001;127(1):71-4.
- Lane W, Zanol K. Duodenal atresia, biliary atresia, and intestinal infarct in truncal aplasia cutis congenita. *Pediatr Dermatol* 2000;17(4):290-2.
- Ribuffo D, Costantini M, Gullo P, Houseman ND, Taylor GI. Aplasia cutis congenita of the scalp, the skull, and the dura. *Scand J Plast Reconstr Surg Hand Surg* 2003;37(3):176-80.
- Kelly BJ, Samolitis NJ, Xie DL, Skidmore RA. Aplasia cutis congenita of the trunk with fetus papyraceus. *Pediatr Dermatol* 2002;19(4):326-9.
- Maccario S, Fasolato V, Brunelli A, Martinelli S. Aplasia cutis congenita: an association with vanishing twin syndrome. *Eur J Dermatol* 2009;19(4):372-4.
- Schaffer JV, Popiolek DA, Orlow SJ. Symmetric truncal aplasia cutis congenita following multifetal reduction of a sextuplet pregnancy. *J Pediatr* 2008;153(6):860-3.
- Simman R. Reconstruction aplasia cutis congenita (group V) of the trunk in a newborn. *Plast Reconstr Surg* 2004;113(3):1103.
- Visva-Lingam S, Jana A, Murray H, John E. Preterm premature rupture of membranes associated with aplasia cutis congenita and fetus papyraceous. *Aust N Z J Obstet Gynaecol*. Feb 1996;36(1):90-1.

Aplasia cutis congenita

26. McCarthy MA, Clarke T, Powell FC. Epidermolysis bullosa and aplasia cutis. *Int J Dermatol* 1991;30(7):481-4.
27. Bigliardi PL, Braschler C, Kuhn P, Sigrist J, Buechner S, Ruffli T. Unilateral aplasia cutis congenita on the leg. *Pediatr Dermatol* 2004;21(4):454-7.
28. Benvenuto C, Kraemer CK, Kruse RL, Cestari TF. Familial epidermolysis bullosa with aplasia cutis congenita: Bart's syndrome?. *Skinmed* 2003;2(5):319-21.
29. Atik B, Tan O, Bayram I, Tuncer O, Kirimi E. Asymmetrical nonscalp aplasia cutis congenita: a case report. *J Dermatol* 2004;31(11):923-6.
30. Boente Mdel C, Frontini Mdel V, Acosta MI, Saleme C, Barrionuevo S, Asial R. Extensive symmetric truncal aplasia cutis congenita without fetus papyraceus or macroscopic evidence of placental abnormalities. *Pediatr Dermatol* 1995;12(3):228-30.
31. Morrell DS, Rubenstein DS, Briggaman RA, Fine JD, Pulkkinen L, Uitto J. Congenital pyloric atresia in a newborn with extensive aplasia cutis congenita and epidermolysis bullosa simplex. *Br J Dermatol* 2000;143(6):1342-3.
32. Izhar R, Ghani T. Aplasia cutis congenita and antithyroid drugs. *J Pak Med Assoc.* Nov 2002;52(11):526-8.
33. Karg E, Bereg E, Gaspar L, Katona M, Turi S. Aplasia cutis congenita after methimazole exposure in utero. *Pediatr Dermatol* 2004;21(4):491-4.
34. Mandel SJ, Brent GA, Larsen PR. Review of antithyroid drug use during pregnancy and report of a case of aplasia cutis. *Thyroid.* Spring 1994;4(1):129-33.
35. Nakamura S, Nishikawa T, Isaji M. Aplasia cutis congenita and skull defects after exposure to methimazole in utero. *Intern Med* 2005;44(11):1202-3.
36. Evers ME, Steijlen PM, Hamel BC. Aplasia cutis congenita and associated disorders: an update. *Clin Genet* 1995;47(6):295-301.
37. Khan JY, Moss C, Roper HP. Aplasia cutis congenita with chromosome 12q abnormality. *Arch Dis Child Fetal Neonatal Ed* 1995;72(3):F205-6.
38. Casanova D, Amar E, Bardot J, Magalon G. Aplasia cutis congenita. Report on 5 family cases involving the scalp. *Eur J Pediatr Surg* 2001;11(4):280-4.
39. Rodrigues RG. Aplasia cutis congenita, congenital heart lesions, and frontonasal cysts in four successive generations. *Clin Genet* 2007;71(6):558-60.
40. Teebi AS, Druker HA. Brachycephaly, cutis aplasia congenita, blue sclerae, hypertelorism, polydactyly, hypoplastic nipples, failure to thrive, and developmental delay: a distinct autosomal recessive syndrome?. *Clin Dysmorphol* 2001;10(1):69-70.
41. Sugiura T, Kouwaki M, Kiyosawa S. A case of systemic aplasia cutis congenita: a newly recognized syndrome?. *Eur J Pediatr.* Apr 2008;167(4):409-13.
42. Canham NL. Cutis aplasia as a feature of Kabuki syndrome. *Clin Dysmorphol* 2006;15(3):179-80.