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CASE REPORT

Extensive aplasia cutis congenita associated with cephalocranial disproportion and brain extrusion

Farbod Torkamand[1] · Aryan Ayati[1] · Zohreh Habibi[1] · Farideh Nejat[1]

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Abstract

Aplasia cutis congenita (ACC) is a rare congenital disorder which can be linked with a variety of other abnormalities. However, the association of this anomaly with cephalocranial disproportion and brain extrusion is rarely reported. In this report, we present a neonate with an extensive ACC with exposed dura mater and sagittal sinus, who later presented with brain extrusion from the defect and an acrocephalic-like feature required decompressive surgery during the first month of life. Theories regarding etiology and progression of acrocephalic feature and brain protrusion in this case have been discussed.

Keywords Aplasia cutis congenita · Cephalocranial disproportion · Intracranial hypertension · Sagittal sinus thrombosis

Introduction

Aplasia cutis congenita (ACC) is a rare congenital disorder, described as the absence of skin layers covering the skull [8, 12]. The defect in skin layers of extremities was first defined in 1767 [5]. The so-called aplasia cutis was further coined to the midline defects that usually occur in the scalp. The condition is caused due to mesodermal failure to close, preventing ectoderm from developing normally [10].

Herein, we describe a neonate with an extensive ACC and exposed brain and sagittal sinus, who later presented with an acrocephalic-like feature and a restricted cranial volume required decompressive surgery during the first month of life.

Case report

A female infant was born with ACC. The prenatal evaluations were unremarkable, and postnatal examinations were normal except for a 10-cm scalp and skull defects over the vertex. Dura mater and the superior sagittal sinus were exposed (Fig. 1a). The patient was initially managed by applying a wet dressing. On the fourth day of life, dura mater ruptured in several points, with the brain starting to be herniated through the defects. The superior sagittal sinus turned black and started bleeding. The patient was controlled hemodynamically and rushed to the operating theater to cover the exposed areas. A T-shaped incision was made in frontal zone, just in front of the scalp defect and behind the hairline, to obtain two vascularized full-thickness scalp flaps bilaterally (Fig. 1b). Duraplasty was performed using pericranium graft. The right-side scalp flap was rotated 90° to cover the whole frontal area and anterior part of the defect, and the left-side flap was reflected backward to cover the posterior part of the defect (Fig. 1c). The first postoperative week was uneventful. During the second week, progressive bulging appeared at vertex causing wound disruption, dural tearing, and extrusion of CSF and necrotic brain (Fig. 1d). Physical examinations revealed an unchanged head size since birthtime, but the head had grown vertically and developed an acrocephalic figure resembling multi-suture craniosynostosis. Brain magnetic resonance images (MRI) showed parenchymal protruding through the defect (Fig. 2a). Considering the small size of skull and vertical growth instead of normal axial growth, a volume-restricted condition of the skull with the reminder sutures approaching each other was supposed. Nonetheless, skull computed tomography (CT) was not performed due to parents' concerns about radiation. Ultrasound assessment confirmed sutures overriding. An elective cranial expansion procedure was scheduled. The scalp flaps were released and endoscopic-assisted bilateral coronal suturectomy was

✉ Zohreh Habibi
z-habibi@sina.tums.ac.ir

[1] Department of Neurosurgery, Children's Medical Center, Tehran University of Medical Sciences, No 62, Qarib St., Keshavarz Blvd., Tehran 1419733151, Iran

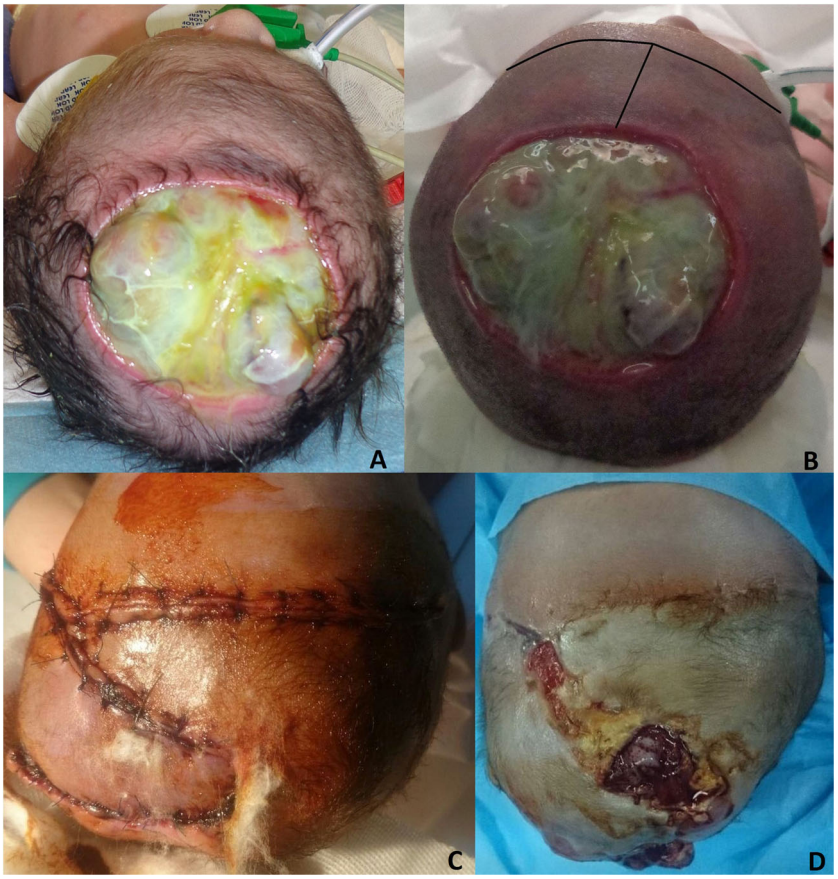

Fig. 1 **a** A photograph shows an extensive aplasia cutis congenita with exposed transparent tiny dura mater and superior sagittal sinus. **b** A T-shaped incision was made in frontal zone, in front of the scalp defect and behind the hairline, to obtain two vascularized full-thickness scalp flaps bilaterally. **c** The right-side flap was rotated to cover the whole frontal area and anterior part of the defect, and the left-side flap was reflected backward to cover the posterior part of the scalp defect. **d** At the end of the second week, progressive bulging appeared at top of vertex causing wound disruption, dural tearing, and extruding CSF and necrotic neural tissue from the wound

performed from the defect toward the base, followed by bilateral lambdoid suturectomy. Areas of dural tearing were repaired, and the skin flaps were replaced to their previous positions (Fig. 2b). Intraoperative observation revealed that the remaining more caudal portion of all four sutures were fused. The postsurgical course was uneventful, and head circumference increased 2 cm postoperatively. The wound healed well, and the skull kept growing normally. After 2 years, the head has normal shape and size. However, the patient suffers from a slight developmental delay and a mild spastic hemiparesis with a left-side porencephalic cyst and ex vacuo ventriculomegaly on MRI (Fig. 2c, d).

Discussion

No definite theory about etiology of ACC has been established. There are several studies showing the importance of genetics [6, 11], or acquired factors like intrauterine vascular compromise and pressure necrosis in amniotic band or pelvic disproportion [13].

The sequels and mortality of ACC are higher in those with extensive exposure of dural sinuses [2]. The small defects can be healed in several weeks without surgical intervention. Wet dressing and close follow-up are used in such cases [7]. Larger defects with extensive dural involvement or hemorrhages from sagittal sinus usually require a kind of surgical repair [2, 8, 12].

ACC could be associated with other abnormalities such as digit anomalies, cleft lip and palate, cerebral malformation, meningomyelocele, frontonasal dysplasia, congenital heart disease, and laryngomalacia [1, 3, 6, 7]. However, the association of this anomaly with cephalocranial disproportion is rarely reported. Only two cases of ACC accompanied with primary craniosynostosis can be found in literature, including one unilateral and one bicoronal synostosis [9, 14]. There is lack of evidence to support the rare association between ACC and either craniosynostosis or secondary cephalocranial disproportion. Secondary cephalocranial disproportion due to cranial sutures overriding and fusion can occur after birth following any type of intracranial hypotension or volume loss, such as overshunting or brain atrophy in early childhood [4, 15].

In the current case, we encountered an acrocephalic feature after ACC repair, resembling the morphology of a multisuture craniosynostosis. Here, two potential hypotheses can be proposed for etiogenesis;

- The first theory is pressure gradient and secondary volume restriction. Considering the course of events, it can be

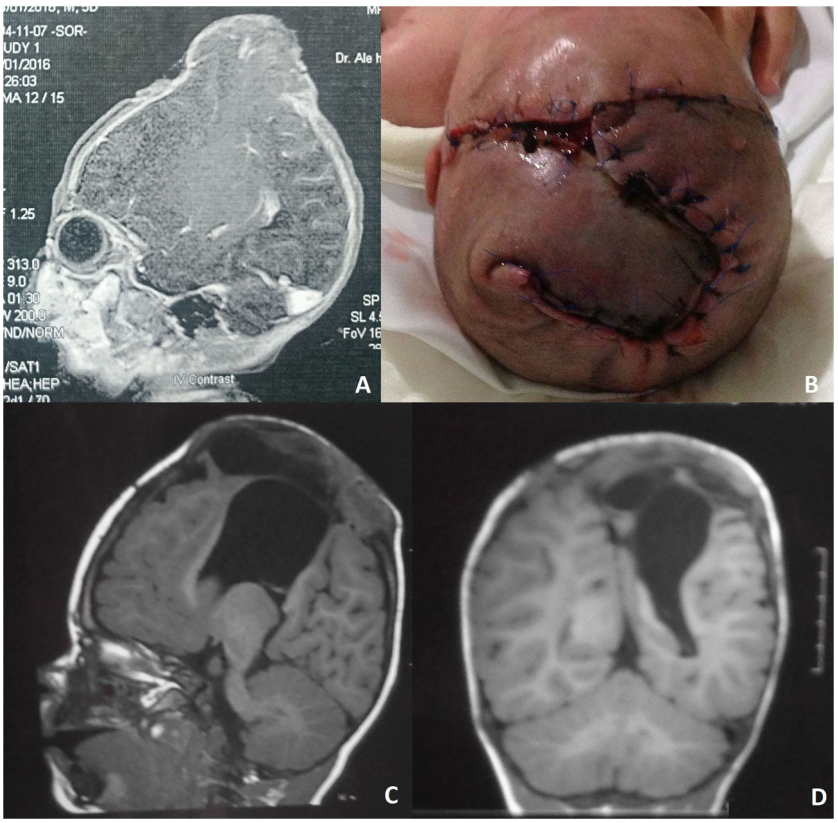

Fig. 2 **a** Brain magnetic resonance images (MRI) showed massive parenchymal protrusion through the bony defect. **b** The skin flaps were replaced to their previous positions and were repaired with the same pattern after bilateral corona and lambdoid suturectomy. **c** and **d**; Follow-up MRI showed a porencephalic cyst due to perinatal brain herniation and necrosis

supposed that brain herniation through the defect had started even before the first surgery, and the initial dural tears and sinus bleeding had occurred in the setting of brain herniation. It can be explained by physical rules of pressure. The intracranial components had been exposed to atmospheric pressure because of the large cranial defect, and the extrusion of brain tissue had been driven by the gradient between the atmospheric and intracranial pressure.

- The second more coherent theory is the role of sagittal sinus involvement and potential venous hypertension due to sinus thrombosis. Considering the dark discoloration of superior sagittal sinus before brain extrusion, it is possible that sinus thrombosis has occurred and caused intracranial hypertension. Ongoing brain herniation would have exacerbated sinus stasis, aggravating intracranial hypertension, and made a vicious cycle. The intracranial volume decreased due to brain extrusion through the boney defect and subsequently caused suture overriding.

Nonetheless, the condition cannot be considered a true secondary craniosynostosis. First, for the reason that 3 weeks is a rather short time to develop secondary fusion. Moreover, as a considerable size of all four sutures was missing, intraoperative observation just confirmed the closure of the remaining most lateral parts of sutures which can be partially fused even in normal neonates. Hence, it is more plausible to assume that the restricted cranial volume was related to brain protrusion due to sinus thrombosis, leading to sutures approaching each other. Also, it seems roughly improbable to assume that the patient had a primary craniosynostosis. None of the genes identified in ACC (including BMS1, KCTD1, ARHGAP31, DOCK6, RBPJ, EOGT, and NOTCH1) [11] are among those involved in craniosynostosis [16].

Conclusion

Extended involvement of superior sagittal sinus, encountered in a very young infant with ACC, is a difficult challenge for neurosurgeons and neonatologists. Surgical treatment is recommended for large ACCs with extensive exposure of dural sinuses. However, there is no consensus about the exact time of surgical repair. Also, there is no evidence about potential protective methods to prevent cerebral sinus thrombosis in these patients. It is theoretically possible that early closure of the defect using skin graft or pedunculated flap may prevent sinus exposure and decrease the risk of thrombosis and subsequent deadly hemorrhagic events. The role of prophylactic anticoagulants has never been investigated in these patients. Providing enough skin coverage for an extensive scalp defect in a so young patient is another challenge that surgical team encounter. Appropriate time and method for cranioplasty is a further dilemma that the therapeutic team may confront as the

child grows up. All abovementioned issues demand further investigations to be answered.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Statement of informed consent Inform consent has obtained from the parents.

References

1. Arand A, Ball W, Crone K (1991) Congenital scalp defects: Adams-Oliver syndrome. Pediatr Neurosurg 17:203–207
2. Burkhead A, Poindexter G, Morrell DS (2009) A case of extensive aplasia cutis congenita with underlying skull defect and central nervous system malformation: discussion of large skin defects, complications, treatment and outcome. J Perinatol 29:582–584
3. Char F, Hansen FC (1971) Frontonasal dysplasia with cutis aplasia congenita. Birth Defects Orig Artic Ser 7(8):265–266
4. Cohen MM (2005) Editorial: perspectives on craniosynostosis. Am J Med Genet A 136A(4):313–326
5. Cordon M (1767) Extrait d'une lettre au sujet de trois enfants de la même mère nés avec partie des extrémités dénuée de peau. J Méd Chir Pharmacie 26:556–557
6. Evers ME, Steijlen PM, Hamel BC (1995) Aplasia cutis congenita and associated disorders: an update. Clin Genet 47:295–301
7. Khashab ME, Rhee ST, Pierce SD, Khashab YE, Nejat F, Fried A (2009) Management of large scalp and skull defects in a severe case of Adams-Oliver syndrome. J Neurosurg Pediatr 4(6):523–527
8. Kim CS, Tatum SA, Rodziewicz G (2001) Scalp aplasia cutis congenita presenting with sagittal sinus hemorrhage. Arch Otolaryngol Head Neck Surg 127:71–74
9. Komuro Y, Yanai A, Seno H, Ichida M, Inoue M, Miyajima M, Arai H, Sato K (2002) Surgical treatment of aplasia cutis congenita of the scalp associated with bilateral coronal synostosis. J Craniofac Surg 13:513–519
10. Kosnik EI, Sayers MP (1975) Congenital scalp defects: aplasia cutis congenita. J Neurosurg 42:32–36
11. Marneros AG (2015) Genetics of aplasia cutis reveal novel regulators of skin morphogenesis. J Invest Dermatol 135(3):666–672
12. Ploplys EA, Muzaffar AR, Gruss JS, Ellenbogen RG (2005) Early composite cranioplasty in infants with severe aplasia cutis congenita: a report of two cases. Cleft Palate Craniofac J 42:442–447
13. Pozzati E, Padovani R, Frank F, Gaist G (1983) Leptomeningeal angiomatosis and aplasia congenita of the scalp: case report. J Neurosurg 58:937–940
14. Spear SL, Mickle JP (1983) Simultaneous cutis aplasia congenita of the scalp and cranial stenosis. Plast Reconstr Surg 71:413–417
15. Weinzweig J, Bartlett SP, Chen JC, Losee J, Sutton L, Duhaime AC, Whitaker LA (2008) Cranial vault expansion in the management of postshunt craniosynostosis and slit ventricle syndrome. Plast Reconstr Surg 122(4):1171–1180
16. Wilkie AO (1997) Craniosynostosis: genes and mechanisms. Hum Mol Genet 6:1647–1656