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Case Report

A Case Of Congenital Aplasia Cutis Occupying A Large Area On The Scalp

Saçlı Deride Geniş Alan Kaplayan Doğuştan Aplazia Kutis Olgusu

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ABSTRACT

Aplasia cutis congenita (ACC) is a rare heterogeneous group of diseases characterized by the absence of skin at birth. It affects males and females equally. It is usually located on the scalp and can be in different sizes from 0.5 up to 15 cm. Typically, it is sporadic, although hereditary cases have also been reported. In cases with bone tissue absence under the lesion, the risk of developing complications increases, and surgical intervention may be required. In this article, we presented a rare case of ACC without underlying bone tissue that covers a very large area on the scalp. In this case, the lesion epithelialized with local treatment without need for any reconstructive surgery.

Keywords: *Aplasia cutis congenita, scalp, wound care*

ÖZET

Aplazia kutis konjenita (AKK) doğumda nadir görülen, cilt yokluğu ile karakterize heterojen hastalık grubudur. Kız erkekleri eşit oranda etkiler. Genellikle skalp yerleşimli olup 0,5 ile 15 cm arasında farklı boyutlarda görülebilir. Tipik olarak sporadik vakalar görülmekle birlikte kalıtsal geçiş gösteren vakalar da bildirilmiştir. Lezyon altında kemik doku olmayan olgularda komplikasyon gelişme riski artmakta, cerrahi müdahale gerekebilmektedir. Bu yazıda, skalpta nadir olarak çok geniş bir şekilde yer kaplayan ve altında kemik dokusu olmayan AKK olgusunu sunduk. Bu olgumuzda herhangi rekonstrüktif cerrahi gerekmeden lokal tedavi ile lezyon epitelize olmuştur.

Keywords: *Aplasia cutis congenita, saçlı deri, yara bakımı*

INTRODUCTION

Aplasia cutis congenita (ACC) is a disease characterized by the absence of skin and subcutaneous tissues at birth. Histopathologically, the epidermis layer is absent or consists of fewer layers. The subcutaneous tissue is thin and has no skin appendages (1). Although sporadic cases are generally seen, hereditary cases have also been reported. Its incidence is 1-3 per 10,000 births, and it is sometimes accompanied by syndromes, such as Johanson-Blizzard and Adams-Oliver (2). ACC is 70 % seen on the scalp and most commonly located on the vertex. It can also be on the trunk and extremities. The size of the defects may range from 0.5 to 15 cm (3,4). In 20% of cases, under the skin defects can be observed in structures, such as bones or dura. The incidence of hemorrhage and infection increases in cases

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accompanied by the absence of bone under the lesion. Surgery may be required in these cases (5).

Different views have been reported on the pathogenesis of the disease in the literature. Prenatal focal ischemia, vascular insufficiency, and prenatal exposure to valproic acid and antithyroid drugs might be the cause of this condition (6,7,8).

A multidisciplinary surgical approach is crucial in the follow-up of ACC cases with scalp lesions, as infections, bleeding, and thrombosis can be seen, and complications, such as seizures, may accompany them. In the case we followed up, there was a 4x5 cm wide aplasia cutis lesion containing immature dura tissue on the vertex region of the scalp, in which the brain sulcus, gyri and sagittal sinus were visible. In this case study, we aimed to share our experience that our patient could be treated with localized wound care under appropriate conditions without the need for surgical treatment in her follow-ups.

CASE REPORT

The patient in this case study was born as the first survivor from the first pregnancy of a 25-year-old mother, at the 37th gestational week, weighing 1990 grams. APGAR was 7/9. The antenatal history of the patient was unremarkable. The mother had not used any medication during pregnancy. The mother did not have a rash disease during pregnancy follow-ups. The baby born by cesarean section had a 4x5 cm wide bone defect on the vertex region of the scalp and a skin defect with clear borders accompanied by the immature dura in which the brain sulci were clearly visible. CSF leakage was not observed. Laboratory examinations were unremarkable. Tests for congenital infections were not significant. Abdominal, cranial USG and ECHO examinations were normal, and respiratory support was not needed in the follow-up. On physical examination, he had hypospadias. Since it may also be associated with Goltz-Gorlin syndrome and some trisomies, karyotype analysis was performed on our patient and the result was 46 XY.

Close follow-up with regular wound dressing was the treatment method we preferred for our patient. Bacitracin 2500 UI and Neomycin 25 mg ointment, which are used for ophthalmic purposes, were used for local wound care for forty days. During the follow-up of our patient, no problems, such as dural rupture, CSF leakage, sepsis, meningitis, bleeding, or skin infections, were encountered. The patient did not require surgical treatment. On the 40th day, the lesion area was reduced, and significant healing was achieved. At the 1-year follow-up, hypertrophic scar tissue developed in our patient, and alopecia developed in the defect area.

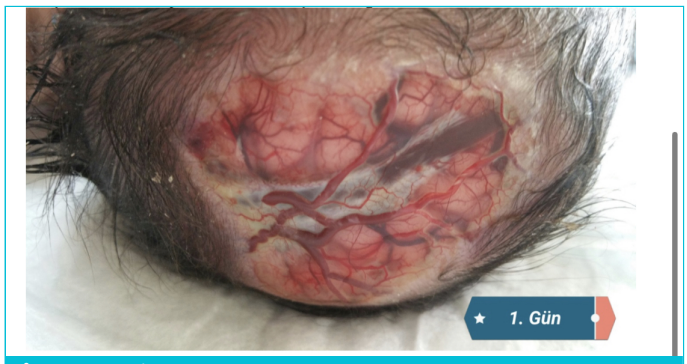

Figure 1. First Day

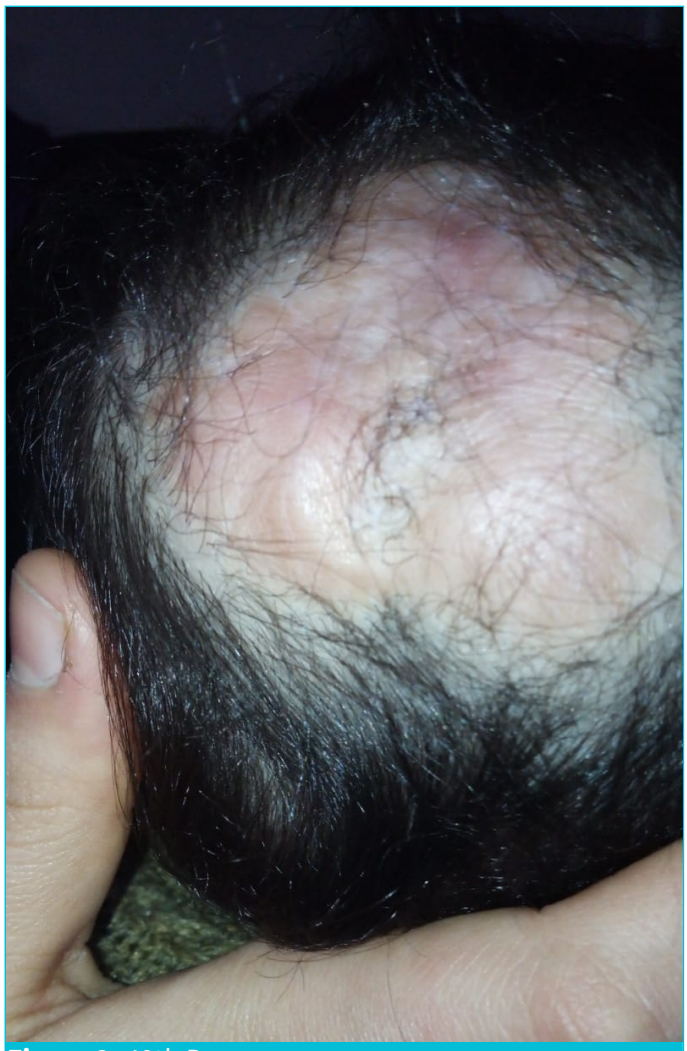

Figure 2. 40th Day

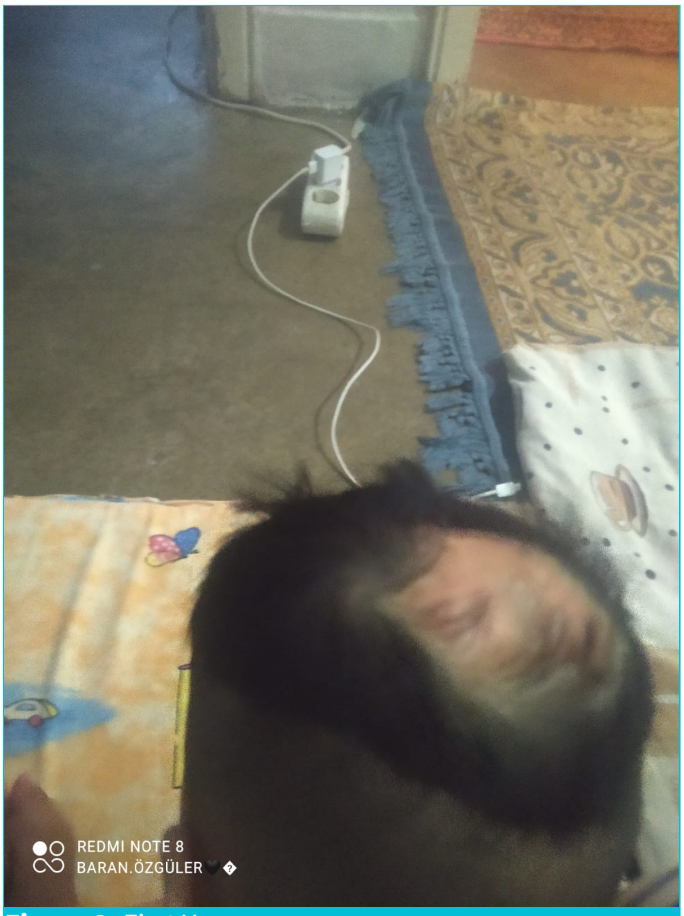

Figure 3. First Year

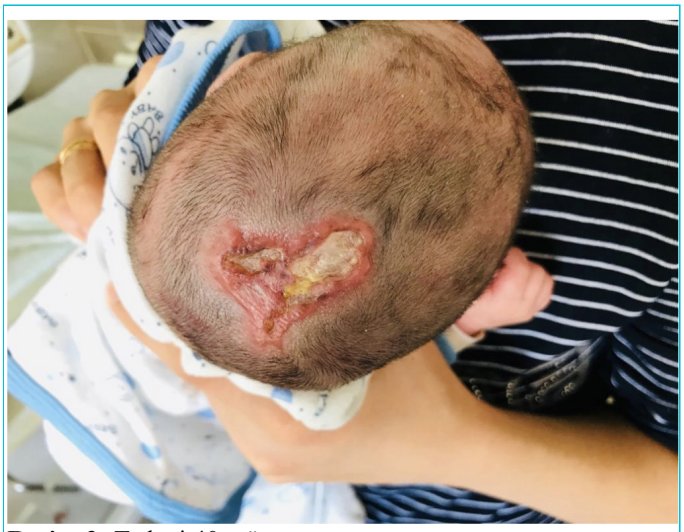

Figure 4. Second Year

DISCUSSION

In conclusion, local care and follow-up of wound healing should be considered before planning surgery at the wound site in patients with ACC. The wound can be left for secondary recovery in patients with ACC who do not have bone and dural defects.

Patient Consent Form / Hasta Onam Formu

The parents' of this patient consent was obtained for this study.

Conflict of Interest / Çıkar Çatışması

The authors declared no conflicts of interest with respect to authorship and/or publication of the article.

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REFERENCES

1. Bajpai M, Pal K. Aplasia cutis cerebri with partial acrania--total reconstruction in a severe case and review of the literature. J Pediatr Surg 2003; 38(2): e4.
2. Marneros AG. BMS1 is mutated in aplasia cutis congenita. PLoS Genet 2013; 9(6): e1003573.
3. Moros Peña M, Labay Matías M, Valle Sánchez F, Valero Adán T, Martín-Calama Valero J, Muñoz Albillos M. Aplasia cutis congenita in a newborn: etiopathogenic review and diagnostic approach. An Esp Pediatr 2000; 52(5): 453-6.
4. Moriya J, Kakeda S, Korogi Y, Soejima Y, Urasaki E, Yokota A. An unusual case of split cord malformation. AJNR Am J Neuroradiol 2006; 27(7): 1562-4.
5. Burkhead A, Poindexter G, Morrell DS. 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 2009; 29(8): 582-4.
6. Hubert A, Bonneau D, Couet D, Berthier M, Oriot D, Larrègue M. Aplasia cutis congenita of the scalp in an infant exposed to valproic acid in utero. Acta Paediatr 1994; 83(7): 789-90.
7. Vogt T, Stolz W, Landthaler M. Aplasia cutis congenita after exposure to methimazole: a causal relationship? Br J Dermatol 1995; 133(6): 994-6.
8. Leboucq N, Montoya y Mártínez P, Montoya-Vigo F, Catan P. Aplasia cutis congenita of the scalp with large underlying skull defect: a case report. Neuroradiology 1994; 36(6): 480-2.