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Aplasia Cutis Congenita of the Scalp, Skull and Dura Associated with Adams-Oliver Syndrome

Konjenital Kafa Derisi, Kemik ve Dura Defektinin Eşlik Ettiği Adams-Oliver Sendromu

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ABSTRACT

A 1-day-old boy with the characteristics of Adams-Oliver syndrome was presented. Adams-Oliver syndrome has a wide spectrum of anomalies ranging from aplasia cutis congenita, cutis marmorata telangiectatica congenita and transverse limb defects to lethal anomalies. Our patient had aplasia cutis congenita with scalp, skull and dura defect. He had also a large dura defect with herniation of brain tissue. Besides these he had bilateral clubfoot, cortical fissure and nail hypoplasia in the hands, scrotal hyperpigmentation and generalized cutis marmorata telangiectatica congenita. He was operated on the 3rd day of life. The herniated brain tissue was resected and the dura was repaired with a synthetic dural graft.

KEY WORDS: Adams-Oliver syndrome, Aplasia cutis congenita, Cutis marmorata

ÖZ

Adams-Oliver sendromlu 1 günlük erkek çocuk vakası sunuldu. Adams-Oliver sendromu konjenital kutis aplazisi, kutis marmaratus telenjiektazi, transvers ekstremite defektleri ve ölümcül anomalilerle karakterize geniş bir spektrum sergilemektedir. Bizim hastamızda konjenital kutis aplazisi, kemik ve dura defekti vardı. Dura defektinden genişçe bir beyin dokusu herniyasyonu vardı. Bunun yanısıra bilateral ayak deformitesi, kortikal fissür, her iki elde tırnak hipoplazisi, skrotal hiperpigmentasyon ve yaygın kutis marmaratus telenjiektazisi mevcuttu. Yaşamının 3. gününde ameliyat edildi. Herniye beyin dokusu rezeke edilerek sentetik dura grefti ile duraplasti yapıldı.

ANAHTAR SÖZCÜKLER: Adams-Oliver sendromu, Kongenital kutis aplazisi, Kutis marmoratus

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INTRODUCTION

Adams-Oliver syndrome (AOS) was first described in 1945. The major components of the syndrome are congenital scalp defects, terminal transverse limb defects and cutis marmorata telangiectatica congenita (CMTC). The syndrome has a wide spectrum of physical anomalies ranging from characteristic defects to extensive lethal anomalies. It was first described as an autosomal dominant disorder but latest reports suggested that it is more commonly sporadic than inherited (1, 5).

Here we report a case of AOS with associated brain anomalies.

CASE REPORT

A full term 1-day-old boy was referred to our hospital with a full thickness scalp and cranium defect. He had also a large dura defect with herniation of brain tissue (Figure 1A). The child's birthweight was 3580 gr, and the APGAR scores at 1 and 5 minutes were 8 and 9. The father was 34 years old and the mother was 28 and they were nonconsanguineous. The mother was treated with an antibiotic which she did not recognize the name for gastroenteritis during her pregnancy.

On physical examination, the child had scalp defect that involved both skin and skull. He also had right and left parietal dura defects separated by an intact superior sagittal sinus. The brain tissue was herniated from the right dura defect. Besides these he had bilateral clubfoot, cortical fissure and nail hypoplasia in the hands, scrotal hyperpigmentation and generalized cutis marmorata telangiectatica congenita (Figure 1B).

An echocardiogram was performed and demonstrated small atrial septal defect (ASD). Magnetic resonance imaging of the brain showed left frontal and parietal multiple ischemic regions. Laboratory examinations were normal except for a sodium of 126 mEq/L (136 – 146 mEq/L) and potassium 2.8 mEq/L (3.4 – 4.5 mEq/L).

He was operated on the 3rd day of life. The herniated brain tissue was resected (Figure 2A) and the dura was repaired by synthetic dural graft (Figure 2B). The skin was closed primarily after the duraplasty.

The patient was discharged 10 days after the operation and he died 2 months after discharge because of pulmonary complications.

DISCUSSION

Since the first description of AOS in eight members of a family, some associated anomalies including brain anomalies, congenital heart disease, ophthalmic abnormalities, spina bifida and intrauterine growth retardation have been reported. Most described cases follow an autosomal dominant pattern of inheritance, but sporadic and autosomal recessive cases have also been reported. Its pathogenesis is unknown. Vascular disruptive mechanisms may be a further sign of vascular disorder as an etiological factor in the development of aplasia cutis congenita, CMTC and AOS (5, 6).

Aplasia cutis congenita is an uncommon disorder with focal absence of epidermis, subcutaneous tissue, galea and calvarial bone in rare cases. Farell et al. reported that 56% of patients with AOS had scalp defects and 21% had skull defects. In AOS, aplasia

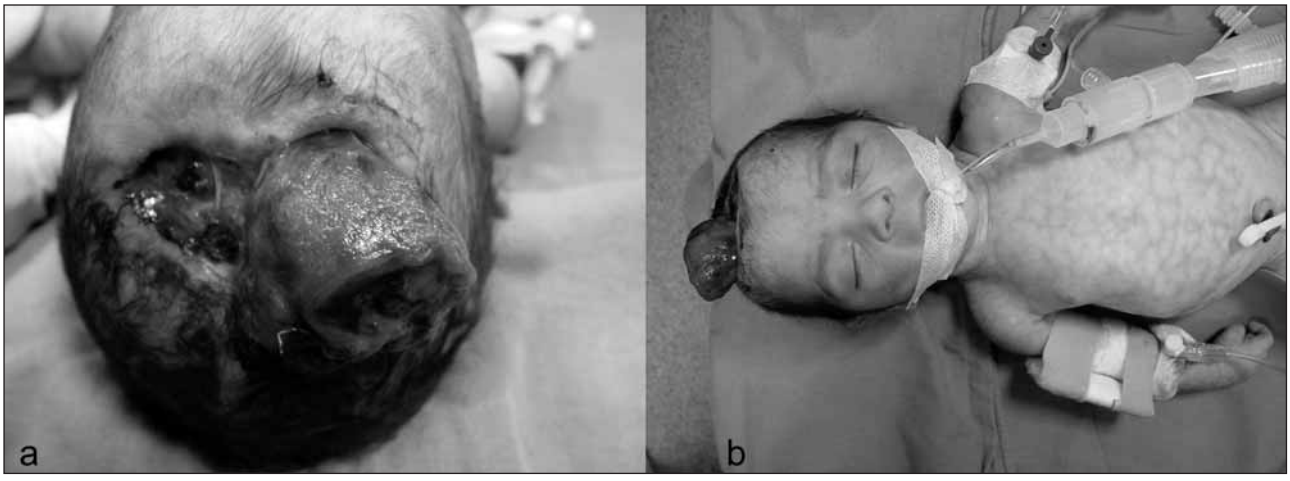

Figure 1: *(**A**) Aplasia cutis congenita with scalp, skull and dura defect, (**B**) Generalized cutis marmorata telangiectatica congenita were seen.*

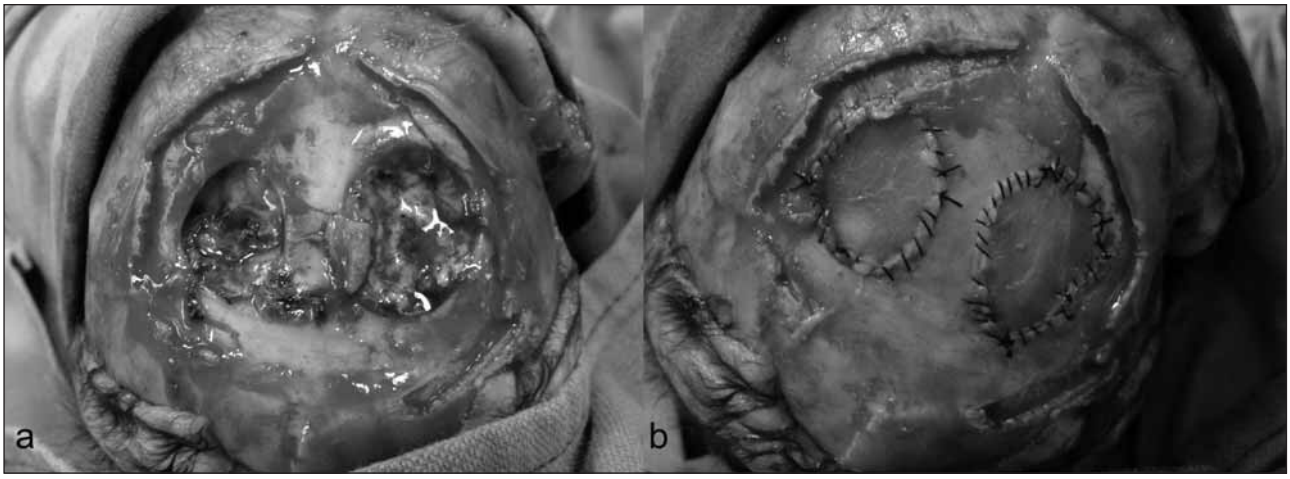

Figure 2: *(**A**) After resection of the herniated brain tissue right and left parietal dural defect with the intact sagittal sinus were seen, (**B**) Duraplasty was performed with a synthetic dural graft.*

cutis congenita generally involves the vertex of the scalp and can be sometimes associated with a bony defect (3). In addition to scalp and bony defect, our patient had a dura defect with herniation of brain tissue.

Characteristically terminal transverse limb defects affect the distal phalanges or entire digits. Both lower and upper limb defects can be seen, but lower limb defects are more common. Shortening of the fingers with loss of terminal phalanges are the most common defects but clubfoot, syndactyly, nail hypoplasia, absence of fingers can be seen less commonly. Limb involvement is usually asymmetrical (3, 5, 6). In our case the patient had bilateral clubfoot, and cortical fissure and nail hypoplasia in the hands.

Cutis marmorata telangiectatica congenita occurs in about 25% of patients. It may be localized or generalized. Its pathogenesis is believed to be a vascular defect (2). Our patient had a generalized type CMTC.

Piazza et al. suggested that CNS abnormalities in AOS patients are consistent with vascular compromise that results in decreased perfusion and ischemia (5). MR imaging of our patient revealed multiple ischemic parietal regions. This finding supports the theory of vascular disruption.

Proper management of aplasia cutis congenita is important. Most cases need conservative treatment including wound dressing with saline and antibiotics. Cases that involve large skin and skull defects like in our patient require surgical management. Mortality rate is about 20% due to infections, meningitis or hemorrhage from the sagittal sinus (4). Early surgical repair of these lesions not only reduces the risk of dural injury but also the mortality rate as well.

We choose early surgical intervention to treat our patient because of the involvement of underlying dura and the brain. After resection of the herniated brain tissue, we performed duraplasty to repair the dural defect with a synthetic dural graft. Some skin defects that cannot be closed primarily may require skin grafting or flaps, but our patient's skin defect closed primarily.

In conclusion, the underlying genetic defect that causes AOS seems to lead to a predisposition for abnormal small vessels causing suspension of blood flow appearing in embryogenesis. Early management of these patients may reduce the risk of developmental delay and mortality because of the vascular pathologies.

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