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An Autopsy Case of Adams-Oliver Syndrome

We report an autopsy case of a male fetus with Adams-Oliver syndrome. His mother was a healthy, 31-year-old woman and her family and past histories were unremarkable. Therapeutic termination was done at 28^{+6} weeks gestational age due to oligohydramnios detected by antenatal ultrasonography. Chromosomal study revealed normal karyotype. On autopsy, characteristic transverse terminal defect of four extremities was found. Both feet were short and broad. All toes were rudimentary with no nails and fingers were irregularly short. On infantogram, all toe-bones were stubby and rudimentary. The middle and terminal phalanges of 2nd, 3rd & 5th fingers and the terminal phalange of 4th finger on the right hand were absent. The middle and terminal phalanges of 2nd & 5th fingers and terminal phalange of 3rd finger were defected on the left hand. His abnormalities were consistent with features of Adams-Oliver syndrome, which has not been reported in Korea.

Key Words: *Adams-Oliver Syndrome; Transverse Terminal Defect of Phalanges*

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INTRODUCTION

"Adams-Oliver Syndrome" is a rare congenital anomaly complex characterized by vertex scalp defect like aplasia cutis congenita, terminal transverse defect of limbs and cutis marmorata of skin. This syndrome is inherited as autosomal dominant with marked variability in expression (1-4). As far as we know, there has been no case reported in Korea.

CASE HISTORY

The male fetus was therapeutically aborted at 28^{+6} weeks gestational age due to oligohydramnios detected by antenatal ultrasonography. The fetus and parents had normal karyotypes on chromosomal analysis. The mother, 31 years old (0-0-3-0), was healthy. She had histories of recurrent abortions, but follow up was not carried out. Her family and past histories were unremarkable.

The baby showed low set ears (Fig. 1). On general inspection, both feet were very short and broad. All toes were rudimentary and no nails were developed. Variable shortness of fingers was also noted but left 4th finger was spared (Fig. 2). On infantogram, all toes-bones did not develop and they were stubby and rudimentary. On the right hand, the middle and terminal phalanges of 2nd, 3rd & 5th fingers and the terminal phalange of 4th finger were absent. On the left hand, the middle and terminal phalanges of 2nd & 5th fingers and terminal phalange of 3rd finger were defected (Fig. 3). Other commonly associated malformations such as scalp defect (aplasia cutis congenita with or without an underlying defect of bone) and skin defect (cutis marmorata) were not demonstrated. No cleft lip and palate were noted (Table 1).

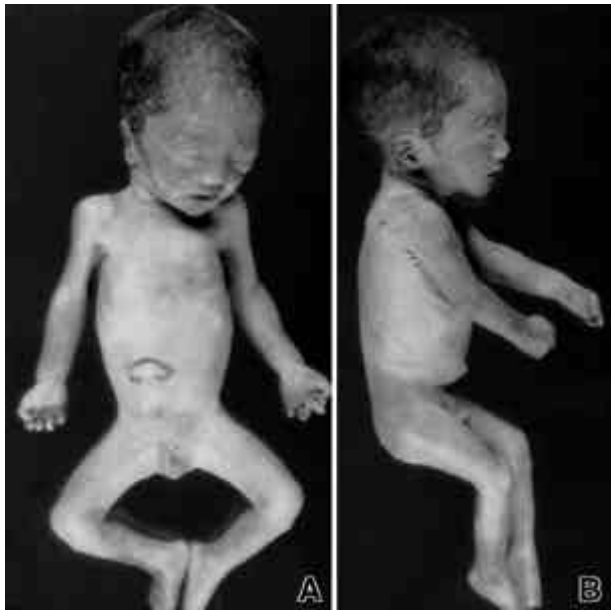

Fig. 1. Prominent abnormalities of both extremities with large head (A) and low set ears (B) are present.

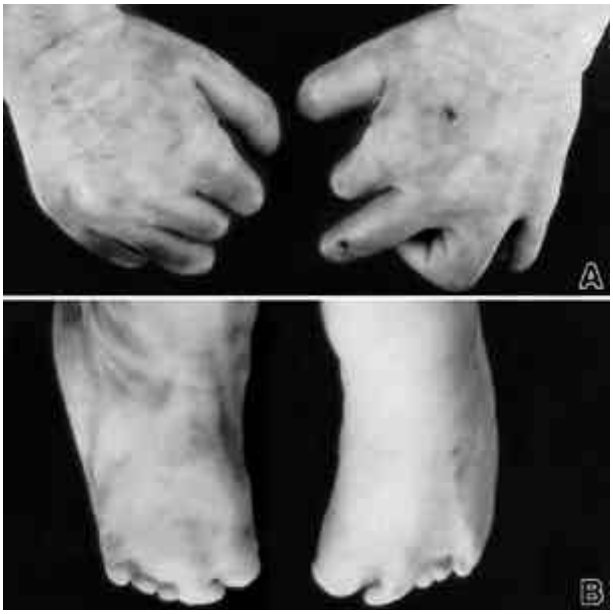

Fig. 2. Variable shortness of fingers (A) and short and broad feet with rudimentary toes.

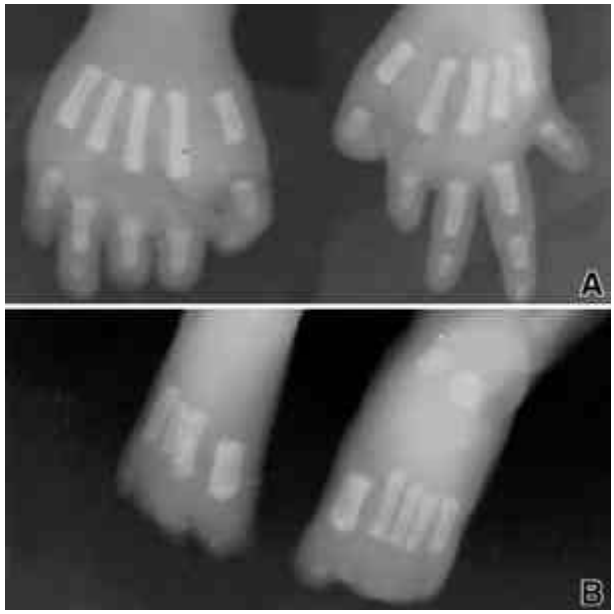

Fig. 3. Postmortem X-ray. Right hand shows defects of middle and terminal phalanges of 2nd, 3rd & 5th fingers and terminal phalange of 4th finger. Left hand shows absence of middle and terminal phalanges of 2nd & 5th fingers and terminal phalange of 3rd finger (A). Rudimentary toes-bones are seen (B).

DISCUSSION

Adams-Oliver syndrome was first described in 1945. Since then more than 40 affected individuals have been reported in the literature, but there has been no case reported in Korea. It is usually inherited as an autosomal dominant trait but autosomal recessive inheritance has been suggested in some families (5).

This syndrome is comprised of defect of terminal phalanges of fingers and toes, aplasia cutis of scalp and cutis marmorata of skin. The scalp lesions reveal thin, atrophic skin or deeper lesions that extend from the skin through the skull to the dura, ranging from 0.5 to 10 cm in diameter (6, 7). Skin and scalp defects were not clearly identified in our case due to his age (28^{+6} weeks gestational age). The terminal transverse limb defect is the most characteristic and major part of this syndrome (4). Limb defects are typically asymmetric and can be more severe in one arm or leg (1, 7). The full spectrum of observed defects ranges from hypoplastic nails, cutaneous syndactyly, bony syndactyly, transverse reduction defects, zygodactyly, ectrodactyly, polydactyly, and brachydactyly (8). More severe defects include complete absence of a hand or foot, or virtual absence of a limb (hemimelia) (1). Our case showed asymmetric involvement of terminal limbs except for the left 4th finger.

Other accompanying anomalies of this syndrome include cryptorchidism, esotropia, accessory nipples, microphthalmia, duplicated collecting system and cleft lip, which were not recognized in our case.

Although our case was too young to observe other abnormalities such as aplasia cutis congenita and cutis

Table 1. Comparison of anomalies of our case and classic Adams-Oliver syndrome

	Features of reported cases	Present case
Limb	Terminal transverse defects (variable expressions)	Terminal transverse defects of fingers and toes (Fig. 2, 3)
Growth	Retarded growth (3rd-10th percentile)	
Skin	Cutis marmorata	Unavailable
Scalp	Aplasia cutis congenita	
Others	Cryptorchidism, Esotropia, Accessory nipples, Microphthalmia, Cleft lip & palate, Duplicated collecting system	Low set ears (Fig. 1)

marmorata, his abnormal features are consistent with Adams-Oliver syndrome.

The pathogenesis of this syndrome is currently unknown (9), but Toriello et al. (10) discussed vascular disruption as the predisposing factor. More cases should be accumulated to better understand this syndrome.

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