Distal Limb Defects and Aplasia Cutis: Adams–Oliver Syndrome

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Adams–Oliver syndrome is a rare congenital condition that should be considered in persons with terminal transverse limb deficiencies and scalp defects (aplasia cutis congenita). Broad phenotypic variability exists in this condition. In its more severe forms, Adams–Oliver syndrome can involve the cardiovascular system, central nervous system, gastrointestinal tract, and genitourinary system and should require prompt evaluation by appropriate subspecialists. Extremity involvement is typically bilateral and asymmetrical, with lower extremities involved more than upper extremities. Brachydactyly is the most common limb defect, and severity ranges from hypoplastic nails to complete absence of the distal limb. The syndrome has been described as resulting from autosomal dominant and recessive modes of inheritance, but most cases are sporadic. No gene has been identified. Although the exact pathogenic mechanism is unknown, a common hypothesis is that a vascular disturbance occurs in watershed areas, such as cranial vertex and limbs, during fetal development. *(J Hand Surg Am. 2016;41(7):e207–e210.*

Key words Adams–Oliver syndrome, brachydactyly, cutis aplasia.

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HAND SURGEONS ARE OFTEN ASKED to evaluate children with distal limb deformities. The immediate concern usually involves addressing the functional needs of the child through appropriate bracing or surgical intervention. Clinicians must be aware that in many cases these deformities may be part of a broader syndrome in which major systemic anomalies may exist that could threaten the child's life. We describe a case of a teenage boy who presented with brachydactyly involving several fingers and toes and an atrophic, hairless patch on the vertex of his scalp. Genetic consultation led to the diagnosis of Adams–Oliver syndrome, a rare syndrome with distal limb deformities that, to our knowledge, has not been presented in the hand surgery literature previously.

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CASE REPORT

A 17-year-old ambidextrous boy presented with terminal transverse deficiencies of all 4 extremities, as well as an atrophic, hairless patch over the vertex of his scalp. He had surgery as a newborn to correct biliary atresia and had excision of a digital remnant of the right great toe at age 4 years.

The boy's scalp defect healed with dressing changes and did not require surgery. He was not found to have other internal organ involvement and his mental and physical development was otherwise normal. His main concern at presentation was snapping of the extensor tendons of the left hand. The right hand had a similar problem but had been corrected surgically a few years earlier.

The boy had been born by normal vaginal delivery at term to nonconsanguineous parents. At the time of the boy's birth, the 22-year-old father was healthy with no apparent physical defects and no known history of congenital anomalies. The 23-year-old mother had reported no problems during the pregnancy and denied

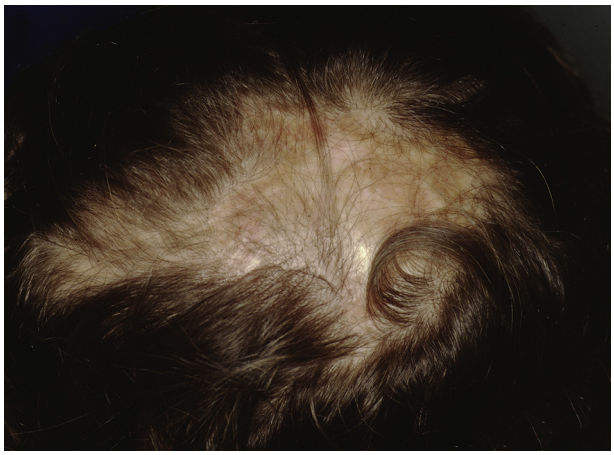

FIGURE 1: Hairless, atrophic patch on the vertex of the scalp (aplasia cutis congenita).

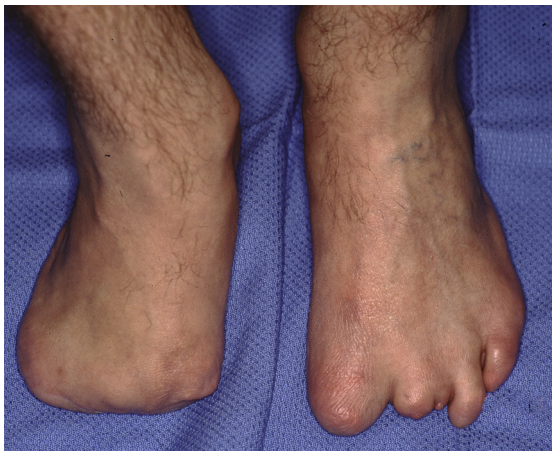

FIGURE 3: Dorsum of both feet of the patient.

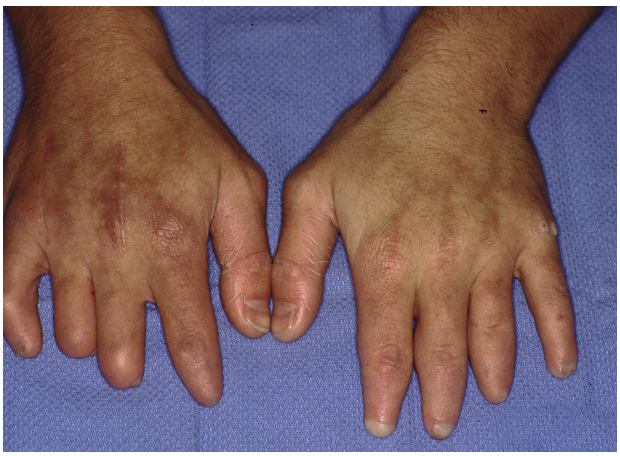

FIGURE 2: Dorsal view of both hands of the patient.

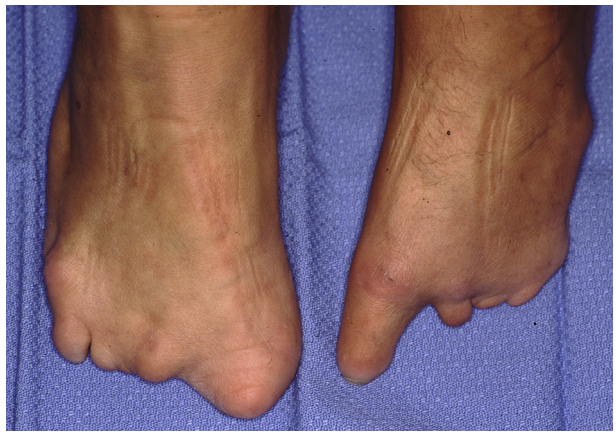

FIGURE 4: Dorsum of both feet of the patient's mother.

use of medications, tobacco, drugs, or alcohol, or exposure to infection or radiation. The boy's younger sister had no skin, skeletal, or limb anomalies and was healthy. The mother claimed that her parents and a sister had no physical anomalies. Her brother had died shortly after birth of a "heart defect." The mother reported being born with terminal deficiencies of the feet and hands and had undergone correction of an atrial septal defect at age 6 years.

On physical examination, the patient appeared healthy without orofacial defects or other obvious signs of dysmorphism. He had a 12 × 5-cm atrophic defect on the scalp (Fig. 1). Hand anomalies included brachydactyly of the ulnar 4 digits on the left hand and the index and little fingers on the right hand. The right middle and ring fingers had transverse terminal deficiencies through the proximal interphalangeal (PIP) joints. He had absent fingernails on the left middle and index fingers and hypoplastic nails on the other digits (except the thumbs). Extension and flexion creases were poorly defined or absent (Fig. 2). Both thumbs were normal in appearance and motion. The patient had full active motion (0° to 100°) at the metacarpophalangeal joints of all digits bilaterally, 0° to 40° active motion at the PIP joints of the right little and index fingers, and 0° to 70° active motion of the other PIP joints. He had painful ulnar subluxation of the extensor digitorum communis tendons involving the ulnar 4 digits of the left hand during composite digital flexion. Evaluation of the boy's feet showed complete loss of the toes on the right and rudimentary hypoplastic toes on the left (symbrachydactyly) (Fig. 3).

The rest of the patient's examination was normal, with no evidence of constriction bands, limited joint mobility, or absent pectoral muscles. His mother agreed to an examination, which showed an atrophic,