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Letter to the Editor

Clinical Evidence of Vascular Abnormalities at Birth in Adams-Oliver Syndrome: Report of Two Further Cases

To the Editor:

We read with interest the recent article by Swartz et al. [1999] on vascular abnormalities in Adams-Oliver syndrome (AOS). Although the pathogenesis of this predominantly autosomal-dominant syndrome remains unclear, it has been suggested [Hoyme et al., 1982] that intrauterine vascular disruption involving small-caliber vessels may be responsible for terminal transverse limb defects. It is possible that vulnerable areas such as the embryonic skin mechanically stretched by rapid brain or limb growth may be affected predominantly by this vascular disruptive mechanism [Pousti and Bartlett, 1997]. According to the hypothesis that interrupted blood supply to the developing structures occurring early in fetal life [Fryns et al., 1996], signs of vascular compromise would not be expected in an infant affected with AOS born at term. The recent admission to our unit of two patients with AOS, one of them with clinical evidence of distal vascular limb compromise at birth, prompts us to report these cases.

Patient 1 was a female born on May 31, 1999, at 36 weeks' gestation after an uncomplicated pregnancy and delivery. Family history was unremarkable. Her birthweight was 2,500 g (25th centile). She was found to have a midline scalp defect of both skin and bone 3 × 5 cm in diameter on the parieto-occipital region, with an intact dura. Her right hand had 2–4 syndactyly and absence of distal phalanges of the 3rd and 5th fingers; her left foot had talipes equinovarus. Her karyotype was normal (46,XX).

Patient 2 was another female born June 6, 1999, at 34 weeks' gestation to a nonconsanguineous 44-year-old mother and a 27-year-old father. Her grandmother was born without nails. Amniocentesis performed for chromosomal analysis showed a normal 46,XX karyotype. Delivery was by cesarean section and birthweight was 1,395 g, length 39 cm, and OFC 26 cm (symmetrical IUGR). An extensive scalp and skull defect was present at birth involving most of parieto-occipital region, with large vascular plaques with atrophy and necrosis; the dura was apparently intact. Her fingertips were necrotic, involving the nails (Fig. 1); the toes were apparently "amputated," and there was evidence of distal ischemia of metatarsal regions (Fig. 2). She also had generalized cutis marmorata telangiectatica, localized ulceration of the abdominal skin, and dilated veins on the trunk. No other malformations were detected. Within 2 weeks after birth, healing of the ischemic le-

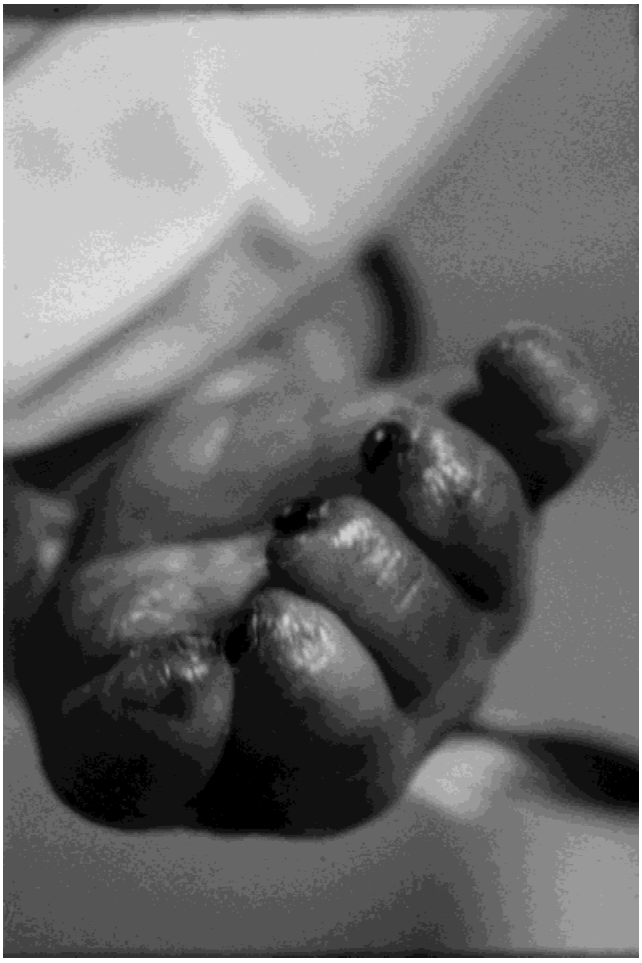

Fig. 1. Congenital necrotic lesions on the fingertips involving the nails.

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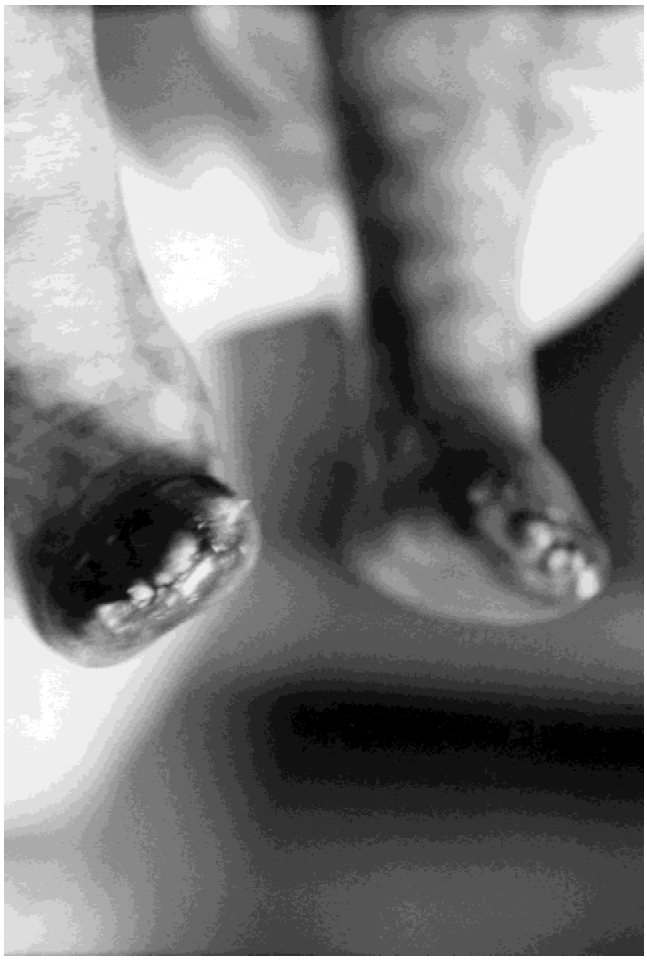

Fig. 2. Absense of toes and distal ischemic lesions on metatarsal regions, present at birth.

sions occurred, the hands and feet stumps became completely covered with skin, and no signs of vascular compromise remained.

We speculate that the clinical evolution of the ischemic lesions observed on the hands and feet in the second case could explain the natural evolution toward the acral deficiencies of AOS during fetal life. In addition, there may also be a possibility of local impairment of circulation occurring later in intrauterine life as well.

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