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J. GORDON MILLICHAP, M.D., F.R.C.P., EDITOR

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CONGENITAL DISORDERS

PENA-SHOKEIR SYNDROME

An infant with Pena-Shokeir syndrome born to a cocaine using mother is reported from the University of Pennsylvania School of Medicine, Philadelphia, PA. The mother had used "\$60 worth of cocaine on the 1st and 2nd of every month" during pregnancy. The Apgars were 2 and 1 and the infant died within minutes after birth. Ultrasound had revealed polyhydramnios in the mother and an autopsy of the infant showed multiple congenital anomalies which included facial anomalies (low-set ears, epicanthic folds, corneal cloudiness, flattened nasal bridge, high-arched cleft palate and micrognathia), skeletal anomalies (arthrogryposis, camptodactyly, ulnar deviation, and bilateral clubfoot), pulmonary hypoplasia, and tetralogy of Fallot with right-sided aortic arch. The brain stem and spinal cord showed diffuse neuronal degeneration and the cerebral cortex showed focal infarction, consistent with acquired intrauterine ischemic damage. The set of anomalies is consistent with the Pena-Shokeir phenotype, also known as the fetal akinesia deformation sequence (Lavi E et al. Fetal akinesia deformation sequence (Pena-Shokeir phenotype) associated with acquired intrauterine brain damage. Neurology Sept 1991; 41:1467-1468).

COMMENT. It was concluded that cocaine-induced vasoconstriction caused ischemic-anoxic brain damage, including damage to motor neurons, and resulted in fetal akinesia deformation sequence. There is a heterogeneity of the neuropathology in previously reported Pena-Shokeir phenotype cases, but most present with congenital CNS malformations caused by neuropathic, myopathic or connective tissue abnormalities that lead to decreased fetal motility in utero.

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