CONGENITAL MALFORMATIONS

BRESEK SYNDROME

Two half brothers with a new syndrome of brain anomalies, retardation, ectodermal dysplasia with alopecia, skeletal malformations, ear abnormalities, and kidney dysplasia (BRESEK) are reported from the University of Minnesota, Minneapolis. The acronym BRESEK denotes the common findings, and BRESHECK reflects additional manifestations occurring in one or other patient: Hirschsprung disease, eye hypoplasia, cleft palate/cryptorchidism. Central nervous system anomalies included microhydrocephaly, dilatation of the spinal canal, and fusion of the thalami. One patient died in respiratory distress at 6 hours. The half brother, maternally related, had neonatal seizures; examination at 7 years of age showed a developmental age of 9 months. Cytogenetic studies in both subjects were normal (46,XY karyotype). (Reish O, Gorlin RJ, Hordinsky M, et al. BRESEK/BRESHECK; new X-linked syndrome. Am J Med Genet Feb 1997;68:386-390). (Respond: Susan A Berry MD, University of Minnesota, Dept of Pediatrics, Box 75 UMHC, 420 Delaware St SE, Minneapolis, MN 55455).

COMMENT. This new syndrome has features similar to some included in syndromes cited by the authors and described by Goldberg and Shprintzen (1981), Brunoni (1983), Santos (1988), Hurst (1988), and Sanyanusin (1995).

CRANIOSYNOSTOSIS PREVALENCE AND TREATMENT

Reasons for the late 1980s apparent epidemic of craniosynostosis and neurosurgical intervention in Colorado, including reports of clusters in selected high-altitude communities, were investigated in the Colorado Department of Health and the University of Colorado School of Medicine, Denver. Of 605 children in the craniosynostosis registry, Colorado Dept of Health, 1986-1989, 307 (51%) had definite radiographic evidence of synostosis. Case reports fell from 347 in the first year to 103 in the third year, following dissemination of information on the controversial nature of the diagnosis and treatment of craniosynostosis. There was also a close surveillance of local diagnostic practices which contributed to the decreased prevalence. Dramatic media coverage of the still unexplained cluster of severe cases in late 1979 preceeded the apparent epidemic. Diagnostic criteria, and the inclusion of radiographically questionable cases, had influenced the rate of synostosis. Evidence for an epidemic of craniosynostosis was not confirmed. (Alderman BW, Fernbach SK, Greene C, Mangione EJ, Ferguson SW. Diagnostic practice and the estimated prevalence of craniosynostosis in Colorado. Arch Pediatr Adolesc Med Feb 1997;151:159-164). (Respond: Beth W Alderman MD, MPH, Department of Epidemiology, University of Washington, Box 357236, Seattle, WA 98195).

COMMENT. CT is more useful than plain radiographs in the diagnosis of coronal and other synostoses. The expert opinion of radiologists is important in diagnostic confirmation prior to surgical intervention.

Diagnosis and management of posterior plagiocephaly is assessed at the Children's Hospital of Pittsburgh, PA. Deformational plagiocephaly was diagnosed in 69 of 71 infants, and only two had true lambdoidal synostosis, associated with posterior sagittal synostosis, and successfully treated surgically. Positional therapy or helmet was generally sufficient. (Pollack IF, Losken HW, Fasick P. Pediatrics Feb 1997;99:180-185).