PEDIATRIC NEUROLOGY BRIEFS
A MONTHLY JOURNAL REVIEW

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

Vol. 11, No. 5 May 1997

PAROXYSMAL NONEPILEPTIC DISORDERS

BREATH-HOLDING SPELLS, ERYTHROBLASTOPENIA, & IRON

An 8-month-old boy with a 1-month history of breath-holding spells and erythroblastopenia is reported from the Naval Medical Center, San Diego, CA. Hemoglobin concentration was 7.6 gm/dl (Hgb A 89.8%, F 7.7%, A2 2.4%), hematocrit 22.6%, reticulocytopenia (0.8%), serum iron 95 mcg/dl, and total iron binding capacity 245 mcg/dl. With iron therapy, breath-holding spells immediately resolved, while the hemoglobin gradually returned to normal after 2 months (Hgb 10.6 g/dl, hematocrit 38.5%). (Tam DA, Rash FC. Breath-holding spells in a patient with transient erythroblastopenia of childhood. J Pediatr April 1997;130:651-653). (Reprints: LCDR DA Tam MC, USN, c/o Clinical Investigation Department, Naval Medical Center, 34800 Bob Wilson Drive, San Diego, CA 92134).

COMMENT. Iron-deficiency anemia and transient erythroblastopenia have both been associated with breath-holding spells and other neurologic and behavioral disorders. The immediate control of breath-holding following iron supplementation, despite persistence of the anemia, is indicative of a specific role of iron in autonomic nervous function.

Effectiveness of iron therapy on breath-holding spells is reported from Jordan University of Science and Technology, Irbid, Jordan (Daoud AS, Batieha A, Al-Sheyyab M, Abuekteish F, Hijazi S. J Pediatr April 1997;130:547-550). Of 33 children treated with ferrous sulfate solution orally (5 mg/kg day for 16 weeks) 88% had a complete or partial control of BHS, whereas in 34 receiving placebo only 6% resolved. Patients with the lower hemoglobin levels at baseline (8.6 gm/dl) had a favorable response whereas those with a mean Hgb 10.6 gm/dl failed to benefit. A low total iron binding capacity was also predictive of a favorable response. Response to iron was correlated with improved blood indexes. Both pallid and cyanotic forms of breath-holding may be associated with iron-deficiency anemia.

Iron deficiency anemia and learning difficulties were correlated in a case-control study of 130 children at Belfast Community

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Paediatric Unit, N Ireland. (Corrigan N, Stewart M, Scott M, Fee F. Fragile X, iron, and neurodevelopmental screening in 8 year old children with mild to moderate learning difficulties. Arch Dis Child March 1997;76:264-267). LD children were more likely to be anemic, had lower serum iron, and failed audiometry tests more frequently than controls. Tests for fragile X, thyroid disorders, and amino acid abnormalities were negative. Iron deficiency anemia and otitis media with hearing impairment should be excluded in the evaluation of children with learning disorders. See Ped Neur Briefs March 1997;11:21, for a report of iron deficiency as a cause of stroke, and Progress in Pediatric Neurology I, Chicago, PNB Publ, 1991;pp397-398, for further reference to iron deficiency and breath-holding, headache, pseudotumor, diplopia, papilledema, and cranial nerve palsies.

NEONATAL HYPEREKPLEXIA

A newborn infant with neonatal sporadic hyperekplexia is reported from the Universita di Napoli Federico II, Italy. Hypertonia and generalized myoclonic jerks after stimulation, noted at 4 days of life, were associated with hypocalcemia. Treatment with calcium gluconate and vitamin D corrected the hypocalcemia, but muscle rigidity and jitteriness persisted. Startle responses were induced by tactile stimuli during sleep but, unlike most cases of hyperekplexia, not in response to tapping the tip of the nose. Hypertonia was relieved and startle responses were milder following treatment with clobazam. At 18 month follow-up, an apnea monitor was discontinued, but clobazam treatment (0.2 mg/kg/daily) was still required. (Scarcella A, Coppola G. Neonatal sporadic hyperekplexia: a rare and often unrecognized entity. Brain Dev April 1997;19:226-228). (Respond: Dr G Coppola, Department of Pediatrics, Universita di Napoli Federico II, Via Pansini, 5, 80131 Napoli, Italy).

COMMENT. Hyperekplexia, or 'startle disease,' of the newborn may occur in major or minor forms, sporadic or familial, with autosomal dominant inheritance, the gene located on chromosome 5q. A myoclonic response to a tap on the nose, hyperreflexia with sustained clonus, hypertonia, jitteriness, and sleep myoclonus are the characteristic manifestations of the major form. The startle response is unaccompanied by stiffness in the minor form. Recurrent apnea, feeding difficulties, choking, and sudden death are reported in some cases.

Packard AM, and Miller VS, of Dallas, TX, report two infants with hyperekplexia who responded to clonazepam (0.05 mg/kg/day), and review 19 reports of 36 families with 220 affected members. (Neurology March 1997;48;A392). Of the cases in the literature, 25 had feeding difficulties in infancy, 21 were apneic, and 10 died in the first year.

Tijssen MAJ et al of Leiden University Hospital, the Netherlands, measured startle reflexes in 9 patients, ages 29 to 66 years, with major hereditary hyperekplexia from the original Dutch pedigree described by Suhren O et al in 1966. (Arch Neurol April 1997;54:388-393). Motor startle responses were more pronounced in patients with a history of major hereditary hyperekplexia than in controls and, contrary to previous reports, also showed more habituation. The authors suggest that cases showing nonhabituation to repetitive stimuli may not fulfill all criteria for the major form of the disorder.