

PEDIATRIC NEUROLOGY BRIEFS

A MONTHLY JOURNAL REVIEW

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

Vol. 2, No. 12

December 1988

ATAXIAS

"HAPPY PUPPET" SYNDROME

The diagnostic features of 36 cases of Angelman's "happy puppet" syndrome are reported from the Hospital for Sick Children, Great Ormond Street, London. These include ataxia, developmental delay, paroxysmal laughter, seizures, and microcephaly. The facial appearance is characterized by a prominent jaw, wide mouth, and pointed chin. The ataxia of gait is associated with jerky limb movements and hand flapping. Muscle tone is variable, deep tendon reflexes normal, and plantar responses flexor. This series included 3 sibships of 3 affected sisters, 2 affected brothers, and 2 affected sisters, respectively. Males and females were equally affected. The electroencephalogram was abnormal in all cases and showed rhythmic slow waves at 4-6HZ and runs of 2-3 HZ anteriorly. On closure of eyes, spikes and 2-4 HZ slow waves occurred posteriorly. CT showed mild cerebral atrophy in 8 of 23 patients tested. Chromosome deletions of 15q11-13 were detected in 5 patients. (Robb SA et al. The "happy puppet" syndrome of Angelman: review of the clinical features. Arch Dis Child Jan. 1989;64:83-86).

COMMENT. Prior to this report, approximately 50 cases of Angelman's syndrome had been described in a period of 20 years. First considered to be sporadic it now appears to be genetic in nature. The authors stress the importance of the recognition of the syndrome so that genetic counseling can be offered.

ASTASIA-ABASIA

Twenty-seven children and adolescents diagnosed as having

PEDIATRIC NEUROLOGY BRIEFS • 1988 covers selected articles from the world literature and is published monthly. Subscription requests (\$28 US or £15 UK annually; add \$5 (£3) for airmail outside North America) may be sent to: Pediatric Neurology Briefs - J. Gordon Millichap, MD, FRCP - Editor, P.O. Box 11391, Chicago, IL 60611, USA, or P.O. Box 1605, London W8 7JD, UK. The Editor, Professor of Neurology and Pediatrics at Northwestern University Medical School, Chicago, is presently at SIU School of Medicine, Springfield, Illinois, USA.