PEDIATRIC NEUROLOGY BRIEFS
A MONTHLY JOURNAL REVIEW

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

Vol. 7, No. 12 December 1993

NEUROENDOCRINE DISORDERS

KALLMANN SYNDROME

The clinical, biological, and molecular genetic aspects of Kallmann syndrome are reported from the Institute of Molecular Genetics, Baylor College of Medicine, Houston, TX. Kallmann syndrome, first described in 1944, is characterized by familial *hypogonadism* and *anosmia* associated with aplasia of olfactory bulbs and tracts. Additional neurologic signs include synkinesia, eye movement abnormalities, horizontal nystagmus, cerebellar ataxia, sensorineural deafness, spastic paraplegia, spatial attention deficits, and mental retardation. Somatic defects including pes cavus, unilateral renal agenesis, and cleft lip and palate are also described. A predominantly X-linked inheritance is reflected in a fivefold excess of male (1 in 10 000) over female (1 in 50 000) patients, and the occurrence of both autosomal dominant and recessive forms indicates genetic heterogeneity. Diagnosis is made at puberty because of a delay in secondary sex characteristics, a eunuchoid habitus, gynecomastia, micropenis, and cryptorchidism. Isolation of the *KAL* gene responsible for the X-linked form of the disease points to a molecular basis for a neuronal migration defect affecting olfactory axons as the primary cause of Kallmann syndrome. (Rugarli EI, Ballabio A. Kallmann Syndrome. From genetics to neurobiology. JAMA Dec 8 1993;270:2713-2716). (Reprints: Dr Rugari, Inst for Molecular Genetics, Baylor College of Med, One Baylor Plaza, Houston, TX 77030).

> **COMMENT.** An adolescent male of eunuchoid build, referred to the neurologist because of attention, coordination and other deficits, should be considered for olfactory tests and an MRI of the olfactory system. Genetics of Kallmann syndrome, see Martin JB. Ann Neurol Dec 1993.

PEDIATRIC NEUROLOGY BRIEFS (ISSN 1043-3155) © 1993 covers selected articles from the world literature and is published monthly. Subscription requests ($43 US; add $12 for airmail outside North America) may be sent to: **Pediatric Neurology Briefs - J. Gordon Millichap, M.D., F.R.C.P.-Editor,** P.O. Box 11391, Chicago, IL 60611, USA. The Editor is Professor Emeritus at Northwestern Univ Medical School and Children's Memorial Hospital. **PNB** is a continuing education service designed to expedite and facilitate current scientific information for physicians and other health professionals.