COMMENT. The syndrome of septo-optic dysplasia appears to be a mild form of holoprosencephaly with single cerebral ventricle and agenesis of the corpus callosum, among other midline defects. Anterior pituitary deficiency is a frequent feature of the syndrome, whereas posterior pituitary disorders are less well documented. In the present study, diabetes insipidus is shown to be a relatively common complication.

CONGENITAL MIDLINE DEFECT IN PITUITARY DWARFS

MRI evaluations of pituitary volume, and clinical and endocrine findings in 101 pituitary dwarfs with congenital idiopathic growth hormone deficiency (CIGHD) are reported from the Departments of Neuroradiology and Pediatrics, Scientific Institute H San Raffaele, Milan, Italy. Ectopia of the posterior pituitary (PPE) was discovered in 59 patients and pituitary volume was reduced. Pituitary hormone deficiency, breech delivery, and other congenital brain anomalies occurred more frequently in PPE patients than in the 42 with normal posterior pituitary except for a narrowed stalk. Associated anomalies included septo-optic dysplasia, with septum pellucidum agenesis and/or hypoplastic optic chiasm, corpus callosum dysgenesis, and basilar impression. A congenital defect involving the pituitary and hypothalamus would account for the MRI abnormalities and the clinico-endocrinological features of CIGHD patients. Breech delivery is the result of the midline brain anomaly, rather than the cause. The hypothesis of a perinatal traumatic transection of the pituitary stalk is contradicted by the findings in this study. (Triulzi F et al. Evidence of a congenital midline brain anomaly in pituitary dwarfs: a magnetic resonance imaging study in 101 patients. Pediatrics March 1994;93:409-416). (Reprints: Dr Fabio Triulzi, Dept of Neuroradiology, Scientific Institute H S Raffaele, via Olgettina 60, 20132 Milano, Italy).

COMMENT. Major brain midline anomalies, including holoprosencephaly, corpus callosum dysgenesis, and septo-optic dysplasia may be associated with hypothalamo-hypophyseal deficiency. Pituitary gland hypoplasia and ectopia, demonstrated by MRI in this and other studies of CIGHD patients, is not correlated with breech delivery, but is related to an anatomical defect in hypothalamic-pituitary structures.

HYDROCEPHALUS IN OSTEOGENESIS IMPERFECTA

The neurological complications of osteogenesis imperfecta in 76 patients are reported from the Human Genetics Branch, National Institute of Child Health and Human Development, NIH, Bethesda, MD. The mean age was 8 years. Communicating hydrocephalus was diagnosed by MRI in 17 patients, macrocephaly in 11, and basilar invagination in 8, with brainstem compression in 3. Seizures occurred in 5 patients, and skull fracture in 10. The importance of detection and treatment of neurological features of osteogenesis imperfecta is noted. (Charnas LR, Marini JC. Communicating hydrocephalus, basilar invagination, and other neurologic features in osteogenesis imperfecta. Neurology Dec 1993;43:2603-2608). (Reprints: Dr Lawrence R Charnas, Building 10, Room 9S242, NIH, Bethesda, MD 20892).

COMMENT. The high frequency of basilar impression in severe cases of osteogenesis imperfecta (OI) was remarkable, in comparison with previous reports. Cervical syringohydromyelia is sometimes a concomitant abnormality with basilar impression.