palate, congenital malformations of the urinary tract. The authors considered that their findings strengthened the hypothesis that BZD intake during early pregnancy is associated with teratogenicity in man but there is as yet no firm proof of association. Diazepam in early pregnancy should be avoided on the basis of this and other studies which strongly suggest a teratogenic effect of BZD.

NEURO-OPHTHALMOLOGY

OCULAR SIGNS OF CHIARI MALFORMATION

Twenty-eight patients (14 females and 14 males aged between four and 34 years) with myelomeningocele and Chiari malformations were examined neuro-ophthalmologically at the Karolinska Institute, Huddinge University Hospital, Huddinge, Sweden. The Chiari malformation, determined with MRI, was type I in three patients and type II in 25. All had hydrocephalus; mild in 12, moderate in 13, and marked in three. Shunt procedures had been performed in 20. Spontaneous or gaze related nystagmus and abnormal optokinetic nystagmus were the most common disturbances of ocular motility. Horizontal gaze paresis occurred in 14 patients and vertical gaze limitations in nine, all in upward gaze. Horizontal nystagmus occurred in 17 whereas vertical nystagmus was uncommon and downbeat nystagmus was not observed. Strabismus occurred in 11 patients and esotropia was more common than exotropia. No signs of optic atrophy or other changes in the visual pathways were found. (Lennerstrand G, Gallo JE. Neuro-ophthalmological evaluation of patients with myelomeningocele and Chiari malformations. Dev Med Child Neur May 1990; 32:415-422).

COMMENT. Downbeat nystagmus has been considered almost pathognomonic of the Chiari malformation but was absent in the cases in this study. A girl aged ten referred to me recently because of migraine headaches had downbeat nystagmus and a refractive error; the MRI was negative for Chiari malformation and brain stem auditory evoked responses were normal. Lennerstrand G et al have used the MRI to determine the correlation between disturbances of ocular motility and the degree of hydrocephalus, tactile plate deformity, and dislocation of the cerebellum and medulla oblongata in 28 patients with myelomeningocele. (Dev Med Child Neur May 1990; 32:423-431). All patients had Chiari malformations. Strabismus and spontaneous nystagmus were related to the degree of hydrocephalus and to the amount of lower brain stem deformities. Convergence defects correlated with upper brain stem deformities.

OPTIC NERVE HYPOPLASIA

The pathology, clinical features, and disorders associated with optic nerve hypoplasia in children are reviewed from the Tennent Institute of Ophthalmology, Weston Infirmary, Glasgow, Scotland. A total of 100 references is provided. Histologically a reduced number of optic nerve fibers can be demonstrated in a smaller than normal optic nerve. An overgrowth of retinal pigment epithelium surrounding

the small optic disc gives rise to the "double ring sign". Failure of differentiation of the retinal ganglion cell layer between the 12 and 17 mm stages of embryonal development has been suggested as a cause of optic nerve hypoplasia (ONH). Other theories include axonal degeneration within the optic nerve or stretching of the optic nerve during the development of abnormal cerebral hemispheres. Factors predisposing to ONH include maternal diabetes mellitus, postmaturity, young maternal age, alcohol abuse during pregnancy, and maternal use of anticonvulsants, quinine, LSD, and phencyclidine. Three clinical varieties are described: 1) Isolated abnormality in an otherwise normal eye; 2) In malformed eyes; 3) With other disorders involving the midline structures of the brain. Nystagmus, poor vision, and visual field defects occur. The differentiation from optic atrophy is important and may require examination with sedation and fundus photography. The electroretinogram is normal but the amplitude of the visual evoked response is reduced. ONH occurs with 25% of cases of agenesis of the septum pellucidum and 27% of patients with ONH have partial or complete absence of the septum pellucidum. The neurological features of this condition, known as septo-optic dysplasia, include mental retardation, spasticity, abnormalities of taste and impaired smell. The ability to learn tasks requiring spatial orientation may be impaired. A number of neurological conditions may be associated with ONH and these include porencephaly, cerebral atrophy, anencephaly, hydrocephaly, congenital suprasellar tumors, and Aicardi syndrome. The frequent association of endocrine problems with ONH should alert the physician to test for pituitary dysfunction early in infancy so that optimal replacement therapy can be given. Children with septo-optic dysplasia and a deficiency of growth hormone frequently have normal growth until their third or fourth year of life. Pituitary dysfunction with ONH may be manifested as diabetes insipidus, prolonged neonatal hyperbilirubinemia, hypotonia, infantile hypoglycemia, hypothyroidism, and growth retardation. All children with ONH should have a careful neuroendocrinology exam including a CT scan. (Zeki SM, Dutton GN. Optic nerve hypoplasia in children. Br J Ophthalmol May 1990, 74:300-304).

COMMENT. The early recognition of optic nerve hypoplasia and its differentiation from optic atrophy are important because of the frequent association with neurological and systemic abnormalities and particularly neuroendocrine disorders which may require early treatment.

NEUROMUSCULAR DISORDERS

JUVENILE AMYOTROPHIC LATERAL SCLEROSIS

Forty-three patients with hereditary motor system diseases belonging to 17 families were studied at the Institut National de Neurologie, La Rabta, Tunis, Tunisia. The mean age of onset was 12 years and the range was three to 25 years. Progression was very slow. Inheritance was autosomal recessive. Patients were subdivided into three groups: 1) Upper limb amyotrophy and pyramidal syndrome (17 patients); 2) spastic paraplegia and peroneal muscular atrophy (14);