Glioblastoma in patients with colorectal cancer has a more favorable prognosis and longer survival than usual. Familial clustering occurs in families with APC, 40% of families having two members with brain tumors. Tumor DNA testing for the incidence of mutations in APC genes in sporadic cases of medulloblastoma would be of interest (Groden J. Colon-cancer genes and brain tumors. N Engl J Med March 30;332:884-5).

OPTIC PATHWAY TUMORS AND PRECOCIOUS PUBERTY

The prevalence of precocious puberty and its relationship to optic pathway tumors (OPTs) have been examined in 219 children with neurofibromatosis 1 (NF-1) seen between Jan 1985 and April 1993 at Children's Memorial Hospital, Chicago. Of seven (3%) with precocious puberty, all had OPTs which involved the optic chiasm and all had abnormal luteinizing hormone-releasing hormone (LH-RH) stimulation tests. They represented 39% of children with NF-1 and chiasmal tumors. Seventy-six percent of OPTs were detected by screening with neuroimaging rather than by clinical symptoms. Biochemical evidence of premature hypothalamic-pituitary-gonadal axis activation may be demonstrated by LH assay, without provocative testing, before overt signs of puberty appear. (Habiby R, Silverman B, Listernick R, Charrow J. Precocious puberty in children with neurofibromatosis 1. J Pediatr March 1995;126:364-7). (Reprints: Joel Charrow MD, Division of Genetics, Children's Memorial Hospital, 2300 Children's Plaza, Chicago, IL 60614).

COMMENT. Precocious puberty associated with neoplastic invasion of the hypothalamus has been reported with gliomas, hamartomas, infundibulomas, and supracellar cysts. Ford FR, in his classic textbook, Diseases of the Nervous System in Infancy, Childhood and Adolescence, 4th ed (Springfield Illinois, CC Thomas, 1960), refers to precocious puberty seen occasionally in cases of tuberous sclerosis and von Recklinghausen's disease. The association with optic pathway tumors and NF-1, emphasized in the present Chicago study, was noted in a 1979 article cited by the authors (Tertsch D et al. Pubertas praecox in neurofibromatosis of the optic chiasm. Acta Neurochir (Wien); 28(suppl):413).

DEGENERATIVE - METABOLIC DISEASES

HARP SYNDROME

Harp syndrome, characterized by hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration, is described in three patients from the National Hospital, Queen Square, London, Newcastle General Hospital, and the Royal Free Hospital, London, UK. An 18-year-old woman presented with intellectual subnormality, night blindness, and dysarthria and dysphagia associated with orobucco-lingual dystonia. T2-weighted MRI showed the "eye-of-the-tiger" sign. This patient's sister and mother had hypoprebetalipoproteinemia but no retinitis pigmentosa or pallidal degeneration. Two patients with a forme-fruste Harp syndrome had the clinical and radiologic features but no lipid abnormality. (Orrell RW et al. Acanthocytosis, retinitis pigmentosa, and pallidal degeneration: A report of three patients, including the second reported case with hypoprebeta-lipoproteinemia (HARP syndrome). (Reprints: Dr Richard W Orrell, Academic Unit of Neuroscience, Charing Cross Hospital, Fulham Palace Rd, London W6 8RF, UK).

COMMENT. HARP syndrome is distinguished from Hallervorden-Spatz disease (HSD) by acanthocytosis and the abnormality of lipoprotein. The authors note that all cases of HARP syndrome have been sporadic and lack the autosomal recessive feature of HSD. For further reports of HARP syndrome, see Progress in Pediatric Neurology II, PNB Publ, 1994, p477.

FRIEDREICH'S ATAXIA WITH RETAINED REFLEXES

Genetic linkage analyses in 11 patients from 6 families with Friedreich's ataxia (FA) phenotype, including cardiomyopathy, but retained reflexes (FARR), are reported from the University of Naples and C Besta Neurological Institute, Milan, Italy; and La Fe University Hospital, Spain. Mean age of onset was 13.5 years. Inheritance was autosomal recessive. All patients had progressive ataxia, dysarthria, dysmetria, scoliosis and pes cavus. FARR mapped to the FA locus on chromosome 9q13-21.1, suggesting that FARR is a variant phenotype of FA. (Palau F et al. Early-onset ataxia with cardiomyopathy and retained tendon reflexes maps to the Friedreich's ataxia locus on chromosome 9q. Ann Neurol March 1995;37:359-362). (Respond: Prof Filla, Clinica Neurologica, Universita Frederico II, via Pansini 5, 80131 Napoli, Italy).

COMMENT. The diagnosis of FARR, a variant of Friedreich's ataxia, should be considered in patients with early onset cerebellar ataxia, cardiomyopathy, and sensory neuropathy. Barbeau found absence of deep tendon reflexes to be a required criterion in the diagnosis of FA (Can J Neurol Sci 1978a;5:57-59), whereas Bell and Carmichael allowed hyperactive reflexes in some cases (Treas Hum Inherit 1939;4:141-281). (Bala V Manyam, personal communication).

INFANTILE LEUKOENCEPHALOPATHY WITH MILD COURSE

Eight children, including 2 siblings, with infantile onset cerebral leukoencephalopathy and megalencephaly, and mild neurological signs and symptoms, are reported from Free University Hospital, and Academic Medical Center, Amsterdam, The Netherlands. Ataxia and spasticity were slowly progressive, while intellectual functioning was preserved for a few years. MRI showed swelling of supratentorial hemispheral white matter, subcortical cysts, and sparing of corpus callosum and internal capsule. Metabolic studies were negative. (van der Knaap MS, Barth PG et al. Leukoencephalopathy with swelling and a discrepantly mild clinical course in eight children. Ann Neurol March 1995;37:324-334). (Respond: Dr MS van der Knaap, Department of Child Neurology, Free University Hospital, PO Box 7057. 1007 MB Amsterdam, The Netherlands).

COMMENT. This type of infantile leukoencephalopathy is distinguished from Canavan and Alexander diseases by an MRI showing severe white matter abnormalities which contrasted with a slow clinical progressive course. Lysosomal and other metabolic white matter disorders characterized by megalencephaly were also ruled out biochemically and clinically.

LATE ONSET KRABBE'S DISEASE WITH PRESERVED INTELLECT in a 24-year-old Swedish male patient is reported from the County Hospital of Jonkoping, and the University of Goteborg, Sweden. (Arvidsson J,