lower limbs are rarely affected at onset in MDS, and the dystonia affects the neck muscles and hands causing neck torsion and writer's cramp. The locus for MDS is mapped to 7q21 region, and 7 different heterogeneous mutations in the gene for E-sarcoglycan (SGCE) have now been identified. An SGCE deficiency is involved in the mechanism of MDS.

NEUROCUTANEOUS SYNDROMES

LATE-ONSET GLIOMA WITH NEUROFIBROMATOSIS TYPE 1

The frequency of symptomatic nonoptic pathway brain tumors in adolescents and adults known to have neurofibromatosis type 1 (NF1) was determined from the National Neurofibromatosis Foundation International Database (NNFFID) in a study at Washington University School of Medicine, St Louis, MO. These tumors were reported in 17 (0.8%) of 2108 patients with NF1 over 10 years of age. The prevalence of brain tumors in patients with NF1 up to 50 years of age is more than 100 times greater than expected, and is significantly elevated at all ages. Of 17 patients with NF1 with adequate clinical information (10 from the NNFFID study and 7 from a National Cancer Institute (NCI) newly diagnosed adult glioma study), brain tumors were diagnosed in 4 cases between 10 and 20 years of age, 7 between 21 and 40 years of age, and in 6 at 41 years or older. Tumors were located in brainstem, cerebellum, cortical and subcortical regions. Three were pilocytic astrocytomas, 6 were grade II, 4 were grade III, and 2 were grade IV (glioblastoma multiforme) neoplasms (grades III and IV in patients older than 20). None had received chemotherapy or radiation previously. Headache was the most frequent presenting symptom (in 15), and paralysis or seizures in 3 patients each. (Gutmann DH, Rasmussen SA, Wolkenstein P et al. Gliomas presenting after age 10 in individuals with neurofibromatosis type 1 (NF1). Neurology September (1 of 2) 2002;59:759-761). (Reprints: Dr David H Gutmann, Department of Neurology, Washington University School of Medicine, Box 8111, 660 S Euclid Ave, St Louis, MO 63110).

COMMENT. Patients with NF1 are at increased risk for the development of astrocytomas beyond 10 years of age and in adult life. In older patients these tumors may be malignant grades III and IV gliomas, not always the more characteristic pilocytic astrocytoma. Population-based epidemiologic studies are recommended, but brain imaging and careful clinical follow-up should be performed in patients with NF1 of any age who present with persistent headache or neurologic abnormalities.

NEUROFIBROMATOSIS TYPE 1 AND SPORADIC OPTIC GLIOMAS

The natural history of sporadic optic gliomas was compared with that of optic gliomas associated with neurofibromatosis type 1 (NF1) in a study using a Children's Tumor Registry (CTR) and an NF1 Database (NF1DB) at St Mary's Hospital, Manchester, UK. A total of 52 cases were identified over a period of 41 years. Of 34 whose natural history was available, 31 were symptomatic. Mean ages of presentation were 4.5 and 5.1 years for NF1 and sporadic cases, respectively. Visual impairment was the presenting complaint in 22, 7 being blind in at least one eye. Recurrence occurred in 12. Overall mortality (47% among NF1 patients and 44% in sporadic cases) and 5 and 10 year survival rates (77% and 67% respectively) were similar in the two groups, but fewer NF1 patients died as a direct result of the optic glioma. Only NF1 optic glioma cases were at risk of developing a second CNS tumor. Five children (29%) with NF1 developed second primary intracranial tumors between 7 and 32 years after initial treatment. Two

had received initial radiotherapy. (Singhal S, Birch JM, Kerr B, Lashford L, Evans DGR. Neurofibromatosis type 1 and sporadic optic gliomas. Arch Dis Child July 2002;87:65-70). (Respond: Dr DGR Evans, Department of Medical Genetics, St Mary's Hospital, Manchester M13 0JH, UK).

COMMENT. Optic gliomas account for less than 5% of childhood brain tumors. The prevalence of neurofibromatosis in patients with optic gliomas in this study is 50% (18/36); estimates range from 10% to 70% in previous cited reports. The prevalence of symptomatic optic glioma occurring in the the above NF1 database patients is 5%. NF1 related optic gliomas are less aggressive and less likely to recur than sporadic cases. Serial eye exams are advised up to 6 years of age, and identified cases should be followed through adult life to rule out development of a second CNS tumor. Sporadic optic gliomas should be treated aggressively, and radiotherapy for NF1 optic gliomas requires clarification.

SEIZURE DISORDERS

STATUS EPILEPTICUS IN CHILDHOOD-ONSET EPILEPSY

The occurrence of status epilepticus, risk factors, and impact on prognosis were determined in a population-based cohort of 150 children (under 16 years old) with new onset epilepsy between 1961 and 1964 and followed prospectively until 1997 at Turku University Hospital, Finland. Forty one (27%) patients developed an episode of status epilepticus (SE), and 22 of these (56%) had 2 or more episodes. Thirty (73%) patients had SE before (12) or at onset (18) of epilepsy and 37 (90%) cases occurred within 2 years of onset. Risk factors for SE included remote symptomatic etiology, age of epilepsy onset at 6 years or younger, abnormal neurologic exam, partial seizures, and specific epilepsy syndromes. Prior febrile seizure was a risk factor for SE with fever and the risk was related to the age of occurrence. SE was not correlated with mortality and affected remission rates only slightly. It did not alter social and educational outcomes. (Sillanpaa M, Shinnar S. Status epilepticus in a population-based cohort with childhood-onset epilepsy in Finland. Ann Neurol September 2002;52:303-310). (Respond: Dr Sillanpaa, Department of Child Neurology, University of Turku Hospital TYKS, 20520 Turku, Finland).

COMMENT. Status epilepticus is a common occurrence in childhood-onset epilepsy. A subgroup of children appears to have a predisposition to SE that occurs early in the course of the epilepsy. The increased risk of SE is correlated with remote symptomatic seizures and younger age of onset. Despite this risk, the long-term prognosis for probability of remission, mortality, and social and educational outcomes is not compromised by the occurrence of SE. Prompt and effective treatment of SE improves the likelihood of a favorable outcome.

MRI findings within 5 days of status epilepticus were studied in 35 children treated at Great Ormond Street Hospital, London, UK. (Scott RC, Gadian DG, King MD et al. Brain September 2002;125:1951-1959). Hippocampal volumes were large in 21 children with prolonged febrile seizures and SE. Patients with afebrile SE had elevated hippocampal T2 values but no hippocampal enlargement. A longitudinal study is required to determine the risk of mesial temporal sclerosis, especially in patients with prolonged febrile seizures and hippocampal edema and enlargement.