off 1.49 MoM). When all measures were included in a discriminant function with stepwise selection of variables, hippocampal height, width of the choroid fissure, and width of the temporal horn alone allowed correct classification of 22 of 26 cases (85% sensitivity) and 20 of 21 controls (95% specificity).

Our data suggest that measures of medial temporal lobe atrophy focusing more precisely on the hippocampus might be useful aids in the diagnosis of Alzheimer's disease even in the early stages of the disease.

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- 1 McKhann G, Drachman D, Folstein MF, Katzman R, Price D, Stadlan EM. Clinical diagnosis of Alzheimer's disease: report of the NINCDS-ADRDA Work Group. *Neurology* 1984;34:939-44.
- 2 Kazee AM, Eskin TA, Lapham LW, et al. Clinicopathologic correlates in Alzheimer Disease: assessment of clinical and pathologic diagnostic criteria. *Alzheimer Dis Assoc Disord* 1993;7:152-64.
- 3 Jobst KA, Smith AD, Szatmari M, et al. Detection in life of confirmed Alzheimer's disease using a simple measurement of medial temporal lobe atrophy by computed tomography. *Lancet* 1992;340:1179-83.
- 4 Scheltens Ph, Leys D, Barkhof F, et al. Atrophy of medial temporal lobes on MRI in "probable" Alzheimer's disease and normal ageing: diagnostic value and neuropsychological correlates. *J Neurol Neurosurg Psychiatry* 1992;55:967-72.
- 5 Binetti G, Magni E, Padovani A, Cappa SF, Bianchetti A, Trabucchi M. Neuropsychological heterogeneity in mild Alzheimer's disease. *Dementia* 1993;4:321-6.

Reversible cortical oedema mimicking cortical dysplasia in mitochondrial disorder

Partial seizures are invariably associated with focal brain pathology. Optimised MRI in the evaluation of these patients for surgery has greatly improved the detection of a spectrum of lesions. Imaging findings, however, are not always specific for a particular pathology, and may transiently mimic a fixed structural lesion.

We briefly report the clinical and laboratory findings of a patient who has had occipital lobe epilepsy since the age of 18. Seizures consisted of frequent and prolonged visual auras (hallucinations, palinopsia, photopsias) followed by reversible but progressive visual loss, as well as complex partial and secondary generalised seizures. Clinical findings were short stature, severe hearing and visual loss, mild ataxia, and dysarthria. The patient's mother had had a stroke at the age of 34 followed by seizures and dementia.

Visual evoked potentials showed abnormal latencies. Monitoring with EEG showed non-specific interictal slowing of background rhythms and focal seizures arising from left and right occipital lobes.

A mitochondrial cytopathy was confirmed by the presence of ragged red fibres and abnormal mitochondrial ultrastructure in the muscle biopsy.

Magnetic resonance imaging during a

Figure 1 Axial T2 weighted MRI obtained during period of increased seizure activity shows widened cortex ribbon due to increased signal of the right occipital cortex and subcortical white matter.

period of increased seizure activity showed thickening of the cortical ribbon of the right parieto-occipital cortex in T1 weighted images. Increased signal was seen in the T2 weighted sequences (fig 1). A diagnosis of cortical dysplasia was considered and the patient was referred to our centre for surgical evaluation. Repeat MRI five months later no longer showed the lesion (fig 2). Retrospectively, it became apparent that the abnormality was due to transient cortical oedema associated with focal status epilepticus and not a fixed structural pathology of the cortex.

Reversible cortical abnormalities have been shown by MRI in generalised and partial status epilepticus.¹⁻³ The appearance may be misdiagnosed as a neoplasm or ischaemic stroke if the transient nature and temporal relation to status epilepticus is not recognised.^{6,7} The unusual linear and pericortical extent of the reversible signal abnormality in our patient led to the initial

Figure 2 Follow up MRI five months later shows resolution of area of focal oedema present in fig 1.

misdiagnosis of a migrational disorder and the patient was referred for evaluation for surgery for epilepsy. Further investigation showed a mitochondrial disorder in our patient and the transient cortical oedema may indeed be secondary to altered cerebral energy metabolism and the pathogenetic mechanisms causing severe seizures in this condition.^{4,5} This case report illustrates that transient functional MRI abnormalities may mimic fixed structural lesions.

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- 1 Callahan D, Noetzel M. Prolonged absence status epilepticus associated with carbamazepine therapy, increased intracranial pressure, and transient MRI abnormalities. *Neurology* 1992;42:2198-201.
- 2 Sammaritano M, Andermann F, Melanson D, et al. Prolonged focal cerebral edema associated with partial status epilepticus. *Epilepsia* 1985;26:334-9.
- 3 Kramer RE, Lüders H, Lesser RP, et al. Transient focal abnormalities of neuroimaging studies during focal status epilepticus. *Epilepsia* 1987;28:528-32.
- 4 Rosen L, Phillips S, Enzmann D. Magnetic resonance imaging in MELAS syndrome. *Neuroradiology* 1990;32:168-71.
- 5 Abe K, Inui T, Hirono N, Mezaki T, Kobayashi Y, Kameyama M. Fluctuating MR images with mitochondrial encephalopathy, lactic acidosis, stroke-like syndrome (MELAS). *Neuroradiology* 1990;32:77.
- 6 Montagna P, Gallassi R, Medori R, et al. MELAS syndrome: characteristic migrainous and epileptic features and maternal transmission. *Neurology* 1988;38:751-4.
- 7 Dvorkin GS, Andermann F, Carpenter S, et al. Classical migraine, intractable epilepsy and multiple strokes: a syndrome related to mitochondrial encephalomyopathy. In: Andermann F, Lugallesi E, eds. *Migraine and epilepsy*. Stoneham: Butterworth Publishers, 1987:203-32.

Absence of SCA1 mutation in idiopathic cerebellar ataxia

Idiopathic cerebellar ataxia refers to a group of sporadically occurring cerebellar degenerations of unknown aetiology, which are clinically characterised by progressive ataxia with an onset in adult life.¹ Neuropathological and clinical studies suggest that there are at least two types of idiopathic cerebellar ataxia. One group of patients presents with additional non-cerebellar symptoms, such as parkinsonism, autonomic failure, and pyramidal symptoms (idiopathic cerebellar ataxia-P). The underlying pathology in many of these patients is olivopontocerebellar atrophy.² Those patients with idiopathic cerebellar ataxia-P who develop severe autonomic failure are subsumed under the broader category of multiple system atrophy.³ The other group of patients is clinically characterised by a pure

cerebellar syndrome. These patients usually have cerebellar cortical atrophy.²

Autosomal dominant cerebellar ataxia resembles idiopathic cerebellar ataxia in many respects. Like idiopathic cerebellar ataxia, autosomal dominant cerebellar ataxia is a late onset disorder that may present as a pure cerebellar disorder or in combination with non-cerebellar symptoms. Neuropathological findings in idiopathic cerebellar ataxia and autosomal dominant cerebellar ataxia are often indistinguishable. These phenomenological similarities have prompted the hypothesis that at least some cases of idiopathic cerebellar ataxia arise from spontaneous mutation of a gene responsible for autosomal dominant cerebellar ataxia, from non-paternity, or falsely appear sporadic due to incomplete pedigree data.^{1,4} The recent discovery of an expanded trinucleotide repeat mutation in the SCA1 region in American families with autosomal dominant cerebellar ataxia offers the opportunity to search for this mutation in isolated patients with idiopathic cerebellar ataxia by the polymerase chain reaction.⁵

We have analysed DNA from 61 patients with idiopathic cerebellar ataxia with a mean age of 53.6 (SD 12.8) years and a mean disease duration of 9.2 (SD 47) years. All patients satisfied the following diagnostic criteria: (a) progressive, otherwise unexplained ataxia, and (b) family history without evidence of heredity or consanguinity of parents. All diagnoses were made after exclusion of possible symptomatic causes (alcoholism, other toxic causes, malignancy, hypothyroidism, vitamin deficiency, inflammatory, metabolic, or vascular causes). Twenty five patients had a pure cerebellar syndrome (idiopathic cerebellar ataxia-C), whereas 36 had additional non-cerebellar symptoms (idiopathic cerebellar ataxia-P). Of these, 17 fulfilled the clinical criteria for probable multiple system atrophy.³ Polymerase chain reaction analysis was performed, as described by Orr *et al.*⁵ For comparison, DNA from 144 patients with autosomal dominant cerebellar ataxia was analysed. As a positive control, DNA from families with autosomal dominant cerebellar ataxia from Yakut tribes from Siberia was included, in which the SCA1 expansion has been demonstrated and sequenced.

A diagnosis of SCA1 heterozygosity was made in five out of 19 German families with autosomal dominant cerebellar ataxia for which the family history was medically documented. This corresponds to a prevalence of SCA1 in German families with autosomal dominant cerebellar ataxia of about 25%. By contrast, the SCA1 expansion was found in none of the 61 cases of idiopathic cerebellar ataxia.

The present results argue against the hypothesis that a relevant number of cases of idiopathic cerebellar ataxia are due to a trinucleotide expansion at the SCA1 locus. The importance of our failure to show a mutation of the SCA1 gene in patients with idiopathic cerebellar ataxia is underlined by the finding that the SCA1 mutation is present in a considerable proportion of German families with autosomal dominant cerebellar ataxia. Our data do not exclude the possibility that some cases of idiopathic cerebellar ataxia are due to spontaneous mutations at other gene loci associated with autosomal dominant cerebellar ataxia. Linkage studies provide evidence that there are at least two other gene loci (SCA2 and SCA3) for autosomal dominant cerebellar

ataxia.^{6,7} The hypothesis that idiopathic cerebellar ataxia is due to a mutation at one of these loci cannot be tested at present because SCA2 and SCA3 genes are not identified. It is unlikely that a recessive gene defect accounts for many cases of idiopathic cerebellar ataxia because recessive inheritance of a late onset ataxic disorder has been documented only very rarely. It therefore seems likely that most cases of idiopathic cerebellar ataxia are non-inherited. This hypothesis is supported by a recent study reporting discordance for idiopathic cerebellar ataxia in monozygous triplets.⁴ In addition, detailed clinical, neuroradiological, and neuropathological investigations show subtle differences between idiopathic cerebellar ataxia and autosomal dominant cerebellar ataxia. Thus age of onset is more variable and tends to be lower in autosomal dominant cerebellar ataxia. Additional non-cerebellar features of idiopathic cerebellar ataxia are often parkinsonism and autonomic failure, whereas ophthalmoplegia, pyramidal signs, dystonia, and amyotrophy are more common in autosomal dominant cerebellar ataxia. Pathological and neuroradiological studies show that there is frequent spinal involvement in autosomal dominant cerebellar ataxia but not in idiopathic cerebellar ataxia.² Finally, postmortem studies have shown the presence of oligodendroglial intracytoplasmic inclusions in the brains of patients with sporadic olivopontocerebellar atrophy, but not in the brains of patients with dominantly inherited olivopontocerebellar atrophy.⁸

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- 1 Harding AE. "Idiopathic" late onset cerebellar ataxia. A clinical and genetic study of 36 cases. *J Neurol Sci* 1981;51:259-71.
- 2 Klockgether T, Wüllner U, Dichgans J, *et al.* Clinical and imaging correlations in inherited ataxias. *Adv Neurol* 1993;61:77-96.
- 3 Quinn N. Multiple system atrophy—the nature of the beast. *J Neurol Neurosurg Psychiatry* 1989;52(special supplement):78-89.
- 4 Harding AE, Sura T, Tippett P, Boughey AM, Patten JP. Discordance for late-onset degenerative ataxia in monozygous triplets. *J Neurol* 1991;238:242-3.
- 5 Orr HT, Chung MY, Banfi S, *et al.* Expansion of an unstable trinucleotide repeat in spinocerebellar ataxia type 1. *Nature Genetics* 1993;4:221-6.
- 6 Gispert S, Twells R, Orozco G, *et al.* Chromosomal assignment of the second (cuban) locus for autosomal dominant cerebellar ataxia (SCA2) to human chromosome 12q23-24.1. *Nature Genetics* 1993;4:295-9.
- 7 Takiyama Y, Nishizawa M, Tanaka H, *et al.* The gene for Machado-Joseph disease maps to human chromosome 14q. *Nature Genetics* 1993;4:300-4.
- 8 Papp MI, Kahn JE, Lantos PL. Glial cytoplasmic inclusions in the CNS of patients with multiple system atrophy (striatonigral degeneration, olivopontocerebellar atrophy, and Shy-Drager syndrome). *J Neurol Sci* 1989;94:79-100.

Life threatening epilepsy in a child

Epilepsy is known to cause a variety of arrhythmias that sometimes lead to sudden death.¹⁻⁴ These arrhythmias are evoked by abnormal autonomic activity to the heart.^{2,3,5} We encountered a patient with epilepsy who developed torsades de pointes. Torsades de pointes—an ECG manifestation in which wide QRS like ventricular tachycardia continuously appears with periodically changing polarity—sometimes leads to ventricular fibrillation, which can cause sudden death.⁶ To our knowledge, this is the first reported incidence of epilepsy that caused torsades de pointes.

A 7 year old girl, who had been investigated for two attacks of epilepsy during the previous six months, was admitted to our institution after respiratory arrest. There was no relevant family history. Her pupils were dilated and there was no light reflex. An ECG showed ventricular fibrillation. After intubation, two attempts at cardioversion with 50 J restored normal sinus rhythm. She was then admitted to the intensive care unit and maintained under controlled ventilation. The ECG taken on admission to the intensive care unit was normal, with no prolongation of the QT interval (QT_c = 0.44 s). About two hours later, a generalised convulsion developed, followed by repeated premature ventricular contractions that immediately changed into torsades de pointes (figure). Xylocaine was ineffective, but diazepam finally terminated both the convulsion and the torsades de pointes. Serum electrolyte concentrations (K 4.5 mmol/l, Ca 9.0 mmol/l, Mg 2.1 mg/dl), and PaO₂ (280 mmHg) were all normal at this time. Thiamylal sodium (3 mg/kg/h) was given for the next three days. During this period the ECG was also normal, with no prolongation of the QT interval. The patient became able to talk two days after barbiturate withdrawal. Myocarditis and cardiomyopathy were ruled out on the basis of echocardiographic findings, and she was negative for all the viral antibodies tested. An EEG obtained in the interictal period was normal. Cranial CT also showed no abnormalities. Thus the arrhythmia was diagnosed as torsades de pointes evoked by generalised seizures. After having been placed on oral anticonvulsant treatment, the patient was transferred to another institution 11 days later without any neurological deficits.

The two points of interest in this case were the induction of torsades de pointes by generalised seizures and the termination of both the seizures and the arrhythmia by diazepam. In this patient, the epileptic focus was not determined, so the attacks were classified as generalised seizures on the basis of their clinical features. As the development of torsades de pointes was preceded by a generalised seizure, the arrhythmia was assumed to be attributable to the seizure. There have been no similar reports. As both conditions developed almost simultaneously, xylocaine was initially given to terminate her torsades de pointes without prolongation of the QT interval, but proved to be ineffective. There was not sufficient time to apply other agents or overdrive pacing. Both conditions were terminated, however, by the diazepam that was subsequently given to control the seizure. Diazepam specifically acts on the limbic system. Considering that arrhythmias secondary to seizures are mediated by a limbic-forebrain-