centres who see all new cases and filter them to the hospital if necessary, and the more traditional hospital-based ones. Vitoria is unusual in that its comparatively large number of neurologists and hospital facilities offers a fully hospital-based neurology service.

We did not study the waiting time: the aim was stated at the beginning of the audit that no patient should wait for more than four weeks and this was checked at the appointment office. This aim was easily achieved and in fact for most of the year the waiting time was around two weeks. No classification as "urgent" or "routine" cases was considered necessary. (In the past, with longer waiting times, we had attempted that classification on the basis of the referring doctor's notes and found it unreliable; we had also tried to briefly interview each patient but it proved impractical). Occasional patients were sent as urgent and seen the same day. Seventy seven per cent of all cases were referred by their general practitioner.

The table shows the main preliminary diagnoses. Sixty six per cent of all new outpatients had conditions that could be termed "non-organic" and 34% had organic problems. The most common diagnoses were headaches (27%), anxiety-depression (19%), dizziness-faints (11%), epilepsy (5%), tremor and other movements (4%), vascular disease (4%).

Five hundred and seventy one patients (58%) were discharged to the referral source; 10 (1%) were admitted to hospital; 139 (14%) were given a further appointment without ordering any tests; 267 (27%) received outpatient investigation, for example, 100 had a CT brain scan, 89 an EEG

and 27 an EMG.

When both outpatient clinics are compared it seems apparent that they are similar. There is a greater percentage of non-organic problems in the Spanish referrals (66%) compared with the English ones (50%) as would be expected from the different settings. But dealing mainly with outpatients who do not have serious disease is probably a universal neurological (medical?) problem which applies even in the restrictive British system.

We would also like to make the following observations:

1) Are scarce resources being wasted with irrelevant problems? The question of who should attend neurological clinics cannot be answered from theoretical principles. We believe it is the system which decides: users' expectations, general practitioners' skills, working conditions and traditions and available resources together produce a net result in the patients we see. It is not clear which variables could be modified to change the pattern: lectures and "educative" discharge letters to general practitioners do not seem to have much effect and increasing or reducing outpatient hours in one English district only changed the number of referrals of the same unselected mix of patients.²

2) Wood *et al*¹ claim that waiting time was "unacceptably long". Here again we feel that acceptability is a relative concept. It would be unacceptable to waste precious time in which an early intervention would have made a prognostic difference, but that hardly seems to be the case in the type of patients seen, as indeed in most non-acute neurology. Once more, acceptability becomes "social acceptability". The British system seems to accept longer waiting times than ours but it would be difficult to prove that your patients are any the worse for it. As an example of the relativity of acceptance, it is often those patients who find waiting for two weeks unacceptable who present themselves with the most variegated arrays of functional symptoms.

3) We read that "there is a considerable shortfall in specialist neurological services in the United Kingdom".¹ Certainly the UK has fewer neurologists than many other countries and a large percentage of neurological problems are dealt with by other physicians. We do not know whether British neurologists generally believe that such problems are badly managed and in fact there seems to be a controversy on the subject.³ But it must be kept in mind that when the number of neurologists reaches a certain point, the question of which patients have to be managed by neurologists ceases to be debatable: neurologists have to deal with all patients traditionally considered neurological. That can considerably change the practice of neurology, with many neurologists becoming a "neurological general practitioner" whose activity largely involves seeing headaches and odd turns in outpatients and strokes on the ward. That can create its own problems such as professional frustration, loss of diagnostic vigilance,⁴ decline of diagnostic and therapeutic skills and a two-tier system with run-of-the-mill neurologists on one side and neurologists of excellence on the other. For many neurologists it may be the end of that seemingly enviable status in which the British neurologist can cope well with his quota of major neurological illness and is provided with "an interesting professional life in the course of which he sees several patients weekly with complex illnesses".⁵

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HLA antigens in familial Guillain-Barré syndrome

The Guillain-Barré syndrome is a sporadically-occurring acute demyelinating polyneuropathy. Its occurrence in first degree relatives is rare with only two reported examples^{1,2} one of which is doubtful.² The search for an association with histocompatibility class I and II antigens (HLA) has been largely unrewarding with no or weak associations.³⁻⁵ We report the occurrence of a Guillain-Barré syndrome in a father and son together with strikingly similar HLA types.

The father was admitted to hospital in 1981 at the age of 58 years one week after a "flu-like" illness. He had a one day history of increasing weakness in the limbs and paraesthesia in the hands, feet and tongue. The tendon reflexes were absent, vibration sense was absent in the feet and he had grade 3/5 weakness proximally in the limbs. The CSF contained protein 300 mg/l and 4 wbc/cmm: oligoclonal bands were present. Facial weakness and ventilatory failure evolved rapidly over three days and he required intermittent positive pressure respiration. He was treated with plasmapheresis. Thereafter, he gradually improved, was discharged two months later and made a complete recovery.

His son was admitted to hospital in 1990 at the age of 43 years two weeks after an upper respiratory infection. There was a five day history of paraesthesia and weakness in the limbs. The tendon reflexes were absent. The CSF protein was raised at 1945 mg/l and there were no oligoclonal bands reported. Nerve conduction studies showed slow conduction in keeping with an acute demyelinating neuropathy. He also deteriorated, was treated with plasmapheresis but did not require ventilatory support. He made a complete recovery over the next three months.

The HLA typing showed remarkably similar results in the father and son. Both had A2, A29, B5, B44, Bw4, Dr7, Drw53. The son's additional results were Dr6, Drw52 and Dqwl.

It is surprising that familial cases have not been described more frequently in a condition in which the immune responses to a variety of viral and other antigenic stimuli may be determined by genetic factors. The HLA types were identical as far as they have been analysed, and they do not include HLA-DR3 or HLA-DR2, both of which have been reported to show weak associations with the

Table New outpatient referrals in 1989

Diagnosis	Number
ORGANIC ILLNESS	337 (34%)
Mental disorders	9
Post-traumatic syndrome	6
Alcoholism	5
Endocrinopathies	4
Sleep disorders	4
Transient global amnesia	2
Degenerative and hereditary	
Tremor and abnormal movements	44
Parkinson's disease	33
Dementia	15
Mental retardation	15
Other degenerative conditions	11
Motor neuron disease	1
Intracranial masses	
Tumour	4
Hydrocephalus	3
Chronic subdural haematoma	1
Other disorders of central nervous system	
Epilepsy	52
Spinal cord disease	6
Multiple sclerosis	1
Disorders of peripheral nervous system	
Bell's palsy	21
Carpal tunnel and other mononeuropathies	20
Root lesions	19
Disorders of other cranial nerves	10
Neuropathy	8
Myopathy	2
Vascular disease	
Stroke	20
Transient ischaemic attack	20
Temporal arteritis	1
NON-ORGANIC ILLNESS	650 (66%)
Headache	265
Anxiety-depression	191
Dizziness-odd turns	56
Syncope	53
Other	69
No disorder	16

disorder.^{4,5} The combination of HLA types in these patients may be a marker for a predisposition to the Guillain-Barré syndrome.

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Adult onset acid maltase deficiency associated with epilepsy and dementia: a case report

Adult onset acid maltase deficiency (AMD) (glycogenosis type II b) characteristically presents after the age of 20 years with a slowly progressive limb-girdle muscle weakness.¹ Respiratory muscle involvement occurs in a third of cases and may be the presenting feature.² Although CNS involvement is seen in the more severe infantile form of AMD (Glycogenosis type IIa), it has not been described in adults. We report a case of adult onset AMD associated with epilepsy and dementia.

A 65 year old woman was referred with intractable epilepsy, the onset of which was at 20 years with a generalised tonic-clonic seizure. Seizures had continued without remission despite treatment with phenobarbitone, phenytoin, and sodium valproate. The current seizure pattern consisted of a secondarily generalised tonic-clonic seizure twice a month and a complex partial seizure approximately monthly. At the age of 29 years she required psychiatric admission because of attempted suicide and hysterical behaviour. She was thought to have an immature personality and to lack foresight and judgement. From the age of 52 years she had been in residential care, and anti-social behaviour, incontinence of urine and impaired cognition had been noted. From the age of 57 years she developed progressive limb weakness.

She had five siblings. One was reported to have epilepsy but the details were not available. Three of the other siblings had adult onset AMD and their histories have been published previously.¹ The parents had no history of any neurological disorder and there was no consanguinity.

On examination cooperation was limited. She was unable to stand. Fundoscopy and eye movements were normal. There was a mild weakness of neck flexion. Wasting of the shoulder girdle muscles and small muscles of the hand was present bilaterally. Wasting was difficult to assess in the legs because of obesity and oedema. There was a grade 4 weakness proximally in the arms, grade 4+ distally, and a grade 2-3 weakness in the legs, more marked proximally. Coordination was

normal. Bilateral grasp reflexes were elicited. Upper limb tendon reflexes were normal but knee and ankle jerks were absent bilaterally. Plantar responses were flexor. No sensory deficit was found. General examination was unremarkable except for obesity.

The following investigations were normal or negative: full blood count, electrolytes, renal function, liver function, thyroid function, serum electrophoresis, autoimmune profile, blood film for acanthocytes, syphilis serology, plasma and urinary amino acids, white cell lysosomal enzymes and nerve conduction studies. The ESR was 45 mm in 1 hour and the creatine kinase was raised at 405 IU/L (0-243 IU/L). Initially serum lactate was slightly elevated at 1.83 mmol/l (0.5-1.65 mmol/l), but was normal on two subsequent occasions. Serum pyruvate was normal. An electrocardiogram showed poor R wave progression but was otherwise normal. Chest radiography revealed a raised right diaphragm. Lying and standing vital capacity were 1.05 l and 1.5 l respectively. Arterial blood gases were normal. During a sleep study lasting 9-18 hours oxygen saturation was 70-80% for 2-3 minutes, 80-90% for 35 minutes and > 90% for the remainder of the study. Electromyographic findings were moderately myopathic without any specific features. Histological appearances of a quadriceps muscle biopsy were indicative of AMD, with a high proportion of the muscle fibres containing multiple vacuoles packed with glycogen. Acid maltase activity in the muscle was reduced at 0.01 (0.07-0.29) μ mol maltose hydrolysed per gram of muscle (wet weight) per minute. Oxidative enzymes, phosphorylase, and adenylate deaminase were normal. The diagnosis was supported by finding a significant number of lymphocytes containing several small discrete glycogen filled vacuoles.³

CT brain scan showed cerebellar atrophy but the patient was unable to comply with an MRI scan. An electroencephalogram showed an irregular dominant rhythm of 8 Hz, widespread theta and delta activity and superimposed bilateral spike and spike wave discharges. Neuropsychological assessment revealed a full scale IQ of 67 (WAIS-R) with evidence of deterioration and most marked impairment of frontal lobe function.

The progressive weakness with evidence of respiratory muscle involvement, muscle histology, and lymphocyte appearances in this patient were all typical of adult onset AMD. Serum lactate was raised on one out of three measurements, which although not a usual feature of AMD may have been secondary to hypoventilation. Additional features in this case were epilepsy, psychiatric disturbance and impaired cognition, for which no other aetiology was identified. Her three siblings with AMD showed a spectrum of clinical and pathological findings,² but did not have epilepsy or dementia, and an elder sister with no history of muscle weakness, had epilepsy. It is possible that, in this family there is a tendency to epilepsy independent of the AMD. An alternative explanation is that the epilepsy, psychiatric disturbance and impaired cognitive function seen in this patient are a manifestation of central AMD.

Although in previous cases of adult onset AMD clinical involvement has been restricted to the skeletal muscles, necropsy studies have revealed more widespread disease,^{4,5} with microscopic abnormalities in all skeletal muscles, whether weak or not, and variable involvement of smooth muscle, cardiac muscle, the tongue and the spleen. In these two

studies of adult onset AMD no microscopic abnormalities were found in the brain.^{4,5} Acid maltase activity is lowest in skeletal muscle, but also reduced in the liver, heart and brain. Cerebral acid maltase activity has been recorded as 6.5% of normal.⁴ There is variation in the degree to which various organs are affected. For example in one post mortem study the level of acid maltase activity in the myocardium was as low as that in skeletal muscle and this was reflected by a vacuolar cardiomyopathy.⁵

In cases of infantile AMD there is more marked reduction of acid maltase activity. Both cardiac and central nervous system manifestations are more prominent, with abnormal glycogen storage in most cells of the brain and spinal cord.

In the few adult onset cases that have been studied, acid maltase activity in the brain has been low.⁴ It is possible that this patient with adult onset AMD has very low levels of cerebral acid maltase activity and that this is the cause of her disturbance of cerebral function. The association of adult onset AMD with epilepsy, cognitive impairment and psychiatric disturbance, however, may be fortuitous and no definite conclusion can be drawn from a single patient. We draw attention to the association in case similar patients have been seen by others.

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A reappraisal of "direction of scratch" test: using somatosensory evoked potentials and vibration perception

We would like to comment on the article by Hankney and Edis¹ which proposed "direction of scratch" test for the clinical examination of posterior column function. Their method is easy to perform, and the errors in the detection of direction of scratch on the skin were reported to be correlated with impaired position and/or vibration sense tested using a 128 Hz tuning fork. Since somatosensory evoked potential (SEP) and vibration sense was thought to be mediated through the posterior column and lemniscal system, we intended to evaluate the usefulness of their test together with SEPs and quantitative vibration sense.

Fifty six patients, mean age 43.5, were studied. They consisted of 43 patients with