disorder. The combination of HLA types in these patients may be a predisposition to the Guillain-Barré syndrome. DLW DAVIDSON AF O'SULLIVAN KD MORLEY Ninewells Hospital and Medical School, Dundee DD1 5SY, UK


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. 

The initial symptom of muscle involvement occurs in a third of cases and may be the presenting feature. 

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 EEG was consistent with 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. 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. Physical examination was unremarkable except for obesity.

The following investigations were normal or negative: full blood count, electrolytes, biochemical function, liver function, renal function, thyroid function, serum electrolysis, autoimmune profile, blood film for acanthocytes, syphilis serology, plasma and urinary amino acids, white cell lysosomal enzymes and nerve conduction studies.

The fatigue was 45 mmol/L at 1 hour and the creatine kinase was raised at 405 IU/L (0-243 IU/L). Initial 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 1-51 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. Electrographic recordings 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 to 0-01 (0-07-0-29) nmol maltose hydrolysed per gram of muscle (wet weight) per minute. Oxidative enzymes, phosphorylase, and adenylyl deaminase were normal. The diagnosis was supported by finding a significant number of lymphocytes containing small, internal discrete glycogen filled vacuoles.

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 92 (WAIS-R) and 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 hyperventilation. Additional features in this case were epilepsy, psychiatric disturbance and impaired cognition, for which no other aetiology was identified. Her three siblings with AMD shared a spectrum of clinical and pathological findings, 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 disease involvement 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.

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. There is variation in the degree to which various organs are affected. For example in one post mortem study the activity in the myocardium was as low as that in skeletal muscle and this was reflected by a vacuolar cardiomyopathy.

In cases of infantile AMD there is more marked involvement with multiple organs and in some cases with central nervous system involvement.

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. 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 found in other disorders and was not drawn from a single patient. We draw attention to the association in case similar patients have been seen by others.

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R reappraisal of “direction of scratch” test: using somatosensory evoked potentials and vibration perception

We would like to comment on the article by Hankney and Edin which proposed “direction of scratch” test for the clinical examination of posterior column function. That 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 128 Hz tuning fork. Since somatosensory evoked potential (SEP) and vibration sense was thought to be mediated through the posterior column and lemniscal pathways, it was used as 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...