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. AMD may have been present 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 phenobarbital, phenytoin, and sodium valproate. The family history consisted of two secondarily generalised tonic-clonic seizures 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.7 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. Visual acuity was unremarkable except for obesity.

The following investigations were normal or negative: full blood count, electrolytes, renal function, liver function, thyroid function, fasting lipids, blood urea, creatinine, urinalysis, urinary calcium/creatine ratio, urinalysis, urine manganese, and urobilinogen. Serum copper, zinc, and ceruloplasmin were normal, as were the IgG, IgA, and IgM levels. Serum angiotensin converting enzyme, 24 hour urinary protein, and a urine protein electrophoresis were normal. Serum albumin and serum cholesterol were normal. Haemoglobin was 13.4 and WBC 4.2 with a differential of 66% neutrophils, 29% lymphocytes, 4% monocytes, and 2% eosinophils. Serum creatine kinase level was 22. Creatine kinase MB isoenzyme was 0.01. Blood glucose, cholesterol, blood urea, electrolytes, and calcium were normal. Urinary amino acids were within normal limits.

On examination of the brain no macroscopic abnormalities were found. There was moderately severe white matter atrophy. On microscopic examination there was evidence of a diffuse axonal loss and astrocytic gliosis. A computerised tomography scan of the head revealed no abnormalities. The clinical and laboratory features excluded multiple sclerosis and paraoxysmal nocturnal hemoglobinuria. A mildly elevated serum creatine kinase level was noted.

The serum creatine kinase level was compared to acid maltase activity in the brain. The acid maltase activity was 0.01. Normal acid maltase activity is 0.05–0.25. Acid maltase activity was therefore reduced to 4–5% of normal.

The clinical and laboratory features were consistent with adult onset AMD. The acid maltase activity in the brain was reduced to 4–5% of normal. The clinical and laboratory features were consistent with adult onset AMD.

We would like to comment on the article by Hankney and Edin5 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 pathway, 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
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