Rare diseases do occur

Sir

Huntingdon’s chorea is an uncommon disease with a worldwide prevalence of 5–10 per 100,000. It usually presents insidiously to a general practitioner, psychiatrist or neurologist and its acute presentation is so rare that it is not to be found in the index of even the largest textbooks on Emergency Medicine. A case is described which first presented to a busy Accident and Emergency Department during a late weekday evening.

The patient was a 44-year-old Jamaican who was brought to the Department by his two employers for whom he had long worked as a gardener. It was explained to the Triage Sister that he was suffering with headaches and ataxia which had become worse that day, but which had actually started after a road traffic accident 2 years previously. The patient was registered at 10.50 pm and a history was later obtained of choreiform movements affecting his arms and legs which had been slowly progressive for no less than 6 years. The employers said that he had become increasingly listless and apathetic in recent months, but the patient himself was unaware of these symptoms and said only that he could not always control the movements of his limbs. He had been treated for syphilis in Jamaica 20 years previously and he said that his mother had a similar problem with her limbs before her early death. He himself was the eldest of seven siblings and he knew that a younger sister was similarly affected. He had two teenage sons who were fit and well. He had indeed suffered an injury to his head following a road traffic accident 2 years previously, but this seemed not to have altered the course of progression of his symptoms.

The most striking feature on clinical examination was the rapid choreiform movement of arms and leg which has accompanied by repeated grimacing of the face and frequent standing on tiptoe. The patient was rather apathetic in appearance, but limited formal testing showed no obvious sign of dimentia. It was after midnight that a presumptive clinical diagnosis was made of Huntingdon’s chorea and this diagnosis was subsequently confirmed by a neurologist following outpatient review. Investigations had revealed immunological signs of treated syphilis and a normal serum copper to exclude Wilson’s Disease, but the diagnosis was confirmed by a CT scan which showed the pathognomonic changes of reduction in volume of the head of the caudate nucleus and of the basal ganglia. Treatment with Tetra-benazine initially improved his chorea and genetic counselling was offered, but declined. One year later he had been lost to follow-up and it will probably be at least 20 years before his sons know whether they are similarly affected. Is this the first case of Huntingdon’s chorea to be diagnosed in an Accident and Emergency Department at midnight?!

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