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A patient that changed my practice

Delayed diagnosis of spinal cord injuries in Huntington's disease

Hamish D Morrison¹, Dafydd Morgan², Duncan J McLauchlan^{3,4}, Catherine Clenaghan³, Anne E Rosser^{3,5}

1. Department of Neurology, Southmead Hospital, Bristol, UK
2. Department of Neurology, University Hospital Wales, Cardiff, UK
3. Division of Psychological Medicine and Clinical Neurosciences, School of Medicine, Cardiff University, Sir Martin Evans Building, Museum Avenue, Cardiff. CF10 3AX
4. Department of Neurology, Swansea Bay University Health Board, Swansea, UK.
5. Brain Repair And Intracranial Neurotherapeutics (BRAIN) Unit, Cardiff University, Hadyn Ellis Building, Maindy Road, Cardiff. CF24 4HQ

Corresponding author: Dr Hamish Morrison, Department of Neurology, Southmead Hospital, Bristol, UK

Contributors: HDM and DM drafted the manuscript and prepared the final submission. DJM, CC and AER undertook patient care and assessment and revised the manuscript.

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Abstract

Huntington's disease is a neurodegenerative disorder, characterised by progressive cognitive, motor and psychiatric symptoms. Patients with advanced disease presenting to emergency medical services can pose a diagnostic and management challenge for physicians unfamiliar with the condition. We describe two patients with Huntington's disease in whom the diagnosis of traumatic spinal cord injury was delayed, and discuss the role cognitive bias played in this delay, and the lessons we can learn.

Introduction

Huntington's disease is a trinucleotide repeat disorder leading to progressive neurodegenerative disease, characterised by motor, cognitive and neuropsychiatric symptoms (Table 1). It is inherited in an autosomal dominant pattern and caused by variable length CAG repeat expansions in the mutated huntingtin gene (mHTT) on chromosome 4.

It is an uncommon disease, with an estimated prevalence of 10.6–13.7 per 100,000 in the Western population. Peak symptom onset is in the fifth decade of life, and it typically progresses over 15–20 years. Its clinical presentation is variable but neuropsychiatric and cognitive problems often precede the motor signs and progression of disease follows a relatively steady trajectory through *early*, *middle* and *late* stage Huntington's disease. Acute deterioration in symptoms is uncommon and should warrant thorough exclusion of an alternative diagnosis.¹

We describe two cases of acute neurological deterioration in patients with Huntington's disease. Both were initially attributed to progression of late stage Huntington's disease but instead proved to be due to traumatic spinal cord injuries.

Table 1 Huntington's disease clinical features

Motor	Chorea, bradykinesia, dystonia, balance/gait disturbance
Cognitive	Deterioration in executive function, impulsivity, lack of insight
Psychiatric	Anxiety, depression, irritability, apathy, perseveration and (more rarely) psychosis

Case 1

A 58-year-old man with a 15-year history of manifest Huntington's disease was admitted to a care home for a period of respite. On review in the Huntington's disease clinic earlier that year he was noted to have significant communication difficulties but could converse, participate in decision making, and walk with assistance. He depended on support for many activities of daily living, primarily due to florid chorea and marked bradykinesia and rigidity. His prognosis at this time was felt to be several years.

After 4 days in respite care his wife received a phone call from the care home informing her that her husband had failed to eat or drink that day and that his head appeared “floppy”, but that care staff were otherwise reassured by the apparent improvement in his chorea. His wife visited the same day and raised concern that he was not using his arms and legs as normal. The next day he developed priapism and could not pass urine. He was taken to the local Emergency Department where urinary retention was confirmed with a residual volume of 800 mL: intermittent urinary catheterisation was initiated and he was discharged. The following day, he had persistent priapism, urinary retention and difficulty passing stool, although care home staff remarked on his calm demeanour despite his difficulties. He reattended the Emergency Department where an indwelling urinary catheter was inserted and he was discharged, despite his wife and care home staff raising the possibility with medical staff of a stroke or alternative diagnosis. A day later a GP attended him in the care home and started antibiotics for a suspected chest or urinary tract infection.

That afternoon the patient was visited by his Huntington’s disease nurse who noted a lack of usual involuntary movements, except for facial chorea. He could not move his limbs, had difficulty with head control and appeared insensate below the neck. On rolling the patient, bruising was noted over his upper spine. The case was discussed with the patient’s neurology team and he was admitted to hospital where routine observations on arrival showed him to be hypothermic, bradycardic and hypotensive. An MR scan of the whole spine identified a cord transection secondary to dislocation of C6/C7 (Figure 1). Operative intervention was precluded by the severity of damage. He died 10 days later in the palliative care unit of a local hospital.

Case 2

A 64-year-old woman with a 15-year history of manifest Huntington’s disease was admitted to her local hospital after a fall at home. Her Huntington’s disease symptoms included a mood disorder with irritability, widespread chorea and mild cognitive impairment. She was independently mobile and able to self-care with no other significant co-morbidities.

On admission she was pyrexial, had raised inflammatory markers with difficulty passing urine and was treated for a urinary tract infection, and subsequently catheterised. She was noted to be “off legs”, attributed to infection on a background of underlying Huntington’s disease. Over subsequent days in hospital she was noted to have progressively deteriorating mobility, with significantly reduced upper limb function. Her family contacted the Huntington’s disease specialist nurse due to their concern that an alternative cause for her weakness has been overlooked. The Huntington’s disease nurse assessment noted a lack of usual involuntary movements except for the face and neck and she requested a medical staff review. At this stage, (11 days into admission) the patient was found to be tetraplegic. An MR scan of spine confirmed spinal cord compression from a posterior subdural haematoma extending from C2 to C4 (Figure 2).

She was transferred to the regional spinal unit and underwent surgical decompression, with subsequent clinical improvement. On discharge from hospital she had not regained purposeful movement although involuntary movements returned after 6 months. She remained wheelchair dependent for mobility, required a long-term urinary catheter and relied on carers for all activities of daily living. She died two years later.

Discussion

These two patients with Huntington's disease developed concerning new neurological problems that warranted further investigation but in both the correct diagnosis was initially overlooked. Each patient presented with features suggesting acute spinal cord pathology. Despite several interactions with healthcare professionals, and family members voicing concern, there was delay in reaching the correct diagnosis until intervention from the Huntington's disease specialist nurse and the clinical deficits became more severe.

In Case 1, the patient's urinary retention was repeatedly dealt with in isolation. The accompanying signs indicating cord compression were not recognised: either missed on examination or mistakenly attributed to pre-existing Huntington's disease. Furthermore, the tetraplegia was misinterpreted as improved chorea (despite there being no reason for expecting chorea to have improved) and provided false reassurance to staff. Although there was no history obtained to support a traumatic injury, identifying the bruising over the spine earlier would likely have hastened diagnostic imaging, although may not have changed the final clinical outcome.

In Case 2, the patient presented to hospital after a fall. The posterior neck soft tissue oedema visible on MR imaging implied a traumatic cause for the cord injury. As there was limb weakness and sphincter dysfunction present from the time of admission, the cord injury probably resulted from this initial fall. Again, there was a lack of a definitive history, and this likely contributed to a delay in diagnosis. Clearly, progressive weakness despite treatment of any underlying infection should prompt re-evaluation of the diagnosis.

Due to cognitive impairment neither patient was able to provide reliable information relating to their acute illness or usual level of function. Additionally, they may have lacked insight into the seriousness of their illness. This is not an uncommon scenario faced by healthcare professionals. Crucial to both cases – emphasised by the intervention from the Huntington's disease nurse – is the need for prior history. What is he/she normally like? What can he/she normally do? Either of these questions would have substantially changed the thinking of the doctors assessing these patients.

Cognitive bias is highly prevalent in medicine, occurring throughout the diagnostic process and estimated to be behind up to 75% of clinical errors². These cases highlight a number of cognitive biases. Firstly, *the framing effect*, whereby positive or negative information that is presented early, in these cases a concurrent diagnosis of Huntington's disease, is overvalued. Secondly, *search satisfying*, whereby clinicians cease to look for further information or alternative explanations when the first plausible solution is found, in these cases urinary retention or urinary tract infection causing a precipitous decline in mobility. Lastly, *diagnostic momentum*, where a clinical course of action instigated by previous clinicians is continued without considering the information available and changing the plan if appropriate, in this case the evolving clinical signs and collateral history from relatives.

Some argue that cognitive biases are inescapable. Whilst the current evidence base for strategies to 'debias' oneself is limited, improving awareness and understanding of our own cognitive bias is a practical first step. Critical thinking i.e. questioning whether the constellation of symptoms and signs could be attributed to Huntington's disease, can be challenging even for experienced clinicians in a time pressured environment, but is essential to avoid misdiagnosis and ensure optimal patient care.

Intercurrent illness is a well recognised cause of deterioration in the clinical features of neurological disease. When reviewing a patient with Huntington's disease experiencing an acute decline in function or behavioural change, one should first consider common and potentially treatable causes¹. Table 2 lists conditions which, in the authors experience, should be considered especially in patients with Huntington's disease.

Table 2 Treatable causes of acute deterioration in patients with Huntington's disease, based on the authors experience

Category	Examples
Infection	Aspiration pneumonia; urinary tract infection, dental abscess; cellulitis
Metabolic	Electrolyte abnormalities; endocrine disorders; nutritional deficiency
Psychiatric	Depression; psychosis
Medication	Adverse effects e.g. neuroleptic malignant syndrome, serotonin syndrome, akathisia; poor adherence
Pain	Early arthritis; occult fracture (hips, wrists)
Other	Subdural haematoma; urinary retention; gastro-oesophageal reflux

Conclusion

We are often reminded that the most important tool in reaching the correct diagnosis is the history, but there is seldom only one story to be heard. Relatives and carers of those with Huntington's disease are often experts and should be consulted along with the usual clinical team wherever possible. Through highlighting these unfortunate cases, we hope that lessons can be learned that improve the care we give to our patients.

Key points

- Huntington's disease has a slowly progressive clinical course and rarely presents with acute severe deteriorations in mobility.
- Obtain a collateral history and be receptive to concerns raised by relatives.
- Be aware of cognitive bias in clinical decision making.
- Effective lines of communication between specialist teams and acute services are essential to ensure optimal care of patients with Huntington's disease.

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