New diagnosis of Huntington disease presenting without chorea
Introduction

Huntington disease is a progressive inherited neurodegenerative disorder

Key features:

- Involuntary movements (chorea)
- Dementia
- Psychiatric symptoms

Huntington disease without chorea has rarely been reported
History

57-year-old left handed female

In her thirties:
- Trouble with social boundaries
- Delusions of ill health
- Diagnosed with schizophrenia aged 36 and treated with risperidone
History

Gradual and slowly progressive deterioration over two to three years
- Cognition
- Gait

Intermittent verbal and physical aggression

Moved to an older adult mental health facility
History

Past medical history
- Hysterectomy for dysmenorrhoea
- Thyroidectomy for benign nodules

Medications
- Lorazepam 0.5 mg twice daily
- Olanzapine 7.5 mg mane, 5mg nocte
- Risperidone 0.5 mg twice daily
- Clonazepam 0.5 mg nocte
- Thyroxine
History

Family history
- Father: died in his 80s of unknown cause; cognitively crisp
- Mother: died from cancer in her 50s; cognitively crisp
- Paternal aunt: schizophrenia
- Older brother: depression with occasional delusions
- Younger brother: died of cancer
- Two younger sisters: cognitively well
- One of her four children reported to have depression
Examination

Formal cognitive examination was not possible

Responded incomprehensively at times; did not follow verbal commands;

Horizontal eye movements appeared intact

Mildly increased tone - neck and limbs

Brisk reflexes
Examination

Gait: slow, wide based and ataxic (required assistance to stand)

No involuntary movements were observed

Pout and bilateral palmo-mental responses
57 F with a neurodegenerative process characterised by cognitive decline and ataxia over 2-3 years on a background of a long term psychiatric disorder
Investigations

MRI brain: generalised atrophy (parietal, temporal, occipital); left predominant; frontal lobes and caudate nuclei relatively spared;

CSF: <1 WCC 80 RBC; protein 0.42 g/L; glucose 3.3;
Diagnosis: Huntington Disease

Genetic testing showed an unstable polymorphic trinucleotide repeat (CAG) in exon 1 of the HTT gene.
Discussion

Huntington disease without chorea has rarely been reported

Case 1:
- 30 M presenting with depressed mood, suicidal ideation and memory decline
- Autopsy
  - No macroscopic atrophy of cortex, basal ganglia, brainstem or cerebellum
  - Immunohistochemistry showed increased reactive astrocytes within the basal ganglia with a reduced number of neurones and increased oligodendrocytes

Discussion

Case 2:
- 42 F presenting with emotional irritability, forgetfulness, and dysarthria
- Central striatum and ventromedial caudate more affected than the putamen and caudate body

Discussion

Long term use of risperidone and olanzapine in our patient would likely have suppressed any chorea.

Dopamine receptor blockers and dopamine depleting agents are often used to treat chorea.

Atypical antipsychotic agents work on a number of receptors including dopamine receptor antagonism.

Important to consider the diagnosis in those with psychiatric symptoms and cognitive decline, particularly if dopamine antagonists are being used.