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Atypical motor presentation of Huntington's Disease

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To the Editors

Huntington's Disease (HD; OMIM# 143100) is a genetic neurodegenerative disorder classically characterised by motor symptoms including chorea, psychiatric and behavioural disturbances, and progressive cognitive deterioration [1]. Chorea, flowing from one body part to another in a non-patterned fashion, is the most characteristic clinical phenomenon in adult-onset HD. On the other hand, patients may also develop an akinetic-rigid syndrome, which is the most common presentation of juvenile HD (known as Westphal variant) in addition to representing the advanced stage of HD when chorea is often no longer present. Less common initial motor presentations of HD exist, e.g. dystonia can be the first noticeable symptom of HD, as well as tics, and cerebellar and pyramidal signs [2].

Here, we describe an uncommon clinical manifestation of HD in a genetically proven case and underline the fact that HD can display a much wider phenotypic spectrum than is usually considered.

A 53-year-old male developed progressive walking difficulties with marked postural instability and balance impairment over a six-month period, in addition to dysarthria, clumsiness in the upper limbs with dystonic posturing, and memory impairment. The patient's grandmother had been affected by a similar condition without a formal diagnosis, his mother had been affected by dementia, and his daughter had a diagnosis of Down's syndrome. On examination, the patient presented with hypomimia, and his eye movements disclosed broken smooth pursuit. He had bilateral upper limb bradykinesia and mild apraxia. Additionally, he presented with generalised dystonia mainly affecting the trunk and upper extremities. His gait was only minimally broad-based, and he was unable to perform tandem gait. Brain MRI revealed prominent putaminal atrophy, and iron deposition in the basal ganglia on T2 sequences, with FC/CC of 1.5 and an increased CC/IT ratio to 0.22 (Fig. 1). The patient was screened for iron storage diseases and Wilson's Disease, both of which came back as negative. Molecular analysis of *HTT* showed a mutant allele with 43 CAG repeats, consistent with a diagnosis of HD. Six months after his initial presentation, he developed generalised chorea.

Here we report a patient with atypical presenting features of HD, namely dystonia. Despite chorea being the most prominent motor symptom of adult-onset HD, recent studies have shed light on non-choreatic movement disorders in HD, either at onset or throughout the disease course [3]. Interestingly, non-choreatic presentations have been correlated with longer CAG repeat expansions and earlier onset [4]. Additionally, MRI has revealed elevated brain iron accumulation (Fig. 1), which had been previously described in HD patients as a result of alteration in metal homeostasis. However, no specific motor presentation has been associated with this brain image [5].

The most common manifestations of HD, other than chorea, seem to be dystonia, ataxia, parkinsonism and tics, although exact numbers are lacking. Parkinsonian signs of HD are less often reported than chorea, but seem to progress

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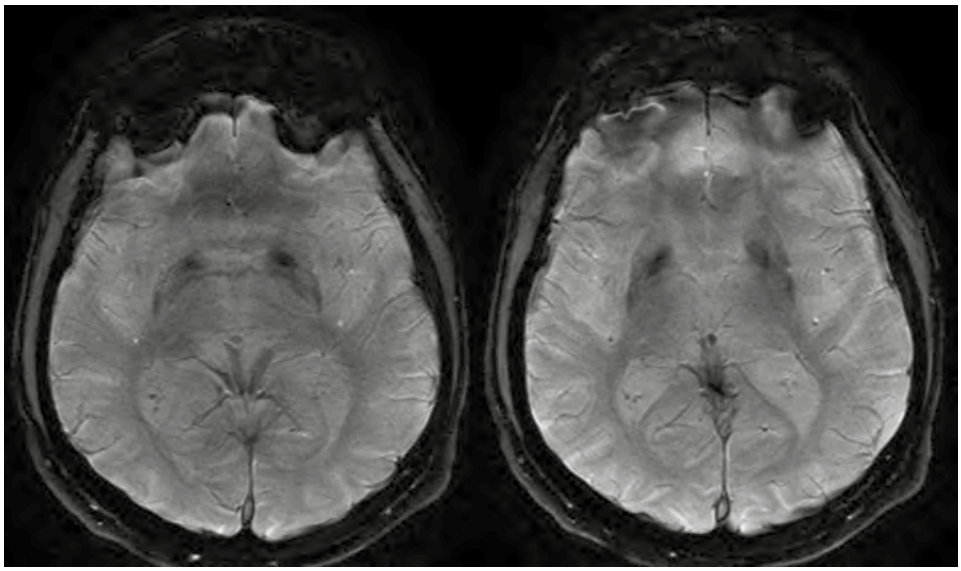

Figure 1. Magnetic Resonance Imaging showing brain iron deposition

in a fairly linear pattern. Becker et al. reported one patient with segmental dystonia, while Hu et al. reported one patient with early-onset blepharospasm followed by cervical dystonia with torticollis and retrocollis; approximately 12% of adult-onset HD patients manifest with prominent dystonia [3].

In conclusion, diagnostic difficulties may arise when HD manifests atypically with non-choreatic motor symptoms. Greater awareness of rare presentations of HD, especially when symptoms overlap with psychiatric co-morbidities, and diligent history-taking regarding the patient's family, will improve diagnosis and aid management. Our case serves to highlight that HD can display a much wider phenotypic spectrum than that which is classically considered, especially in extending to symptoms beyond chorea.

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