

## Huntington's Disease: An Unanticipated Diagnosis

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**Chorea implies erratic, brisk, non-suppressible spontaneous movements mainly of the distal limbs and face. The most common cause of chorea is Huntington's disease, which is a genetic condition affecting the central nervous system.**

**We present a patient with worsening choreiform movements, imbalance, slurred speech, behavioral changes and cognitive decline. His symptoms were initially attributed to a stroke, but eventually led to a diagnosis of Huntington's disease. Currently, there are no cases reported where Huntington's disease was mistaken for a stroke.**

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Chorea implies erratic brisk, non-suppressible spontaneous movements mainly of the distal limbs and face. The most common cause of chorea in the elderly is Huntington's disease, which is a genetic condition affecting the central nervous system<sup>1</sup>. It has an autosomal dominant penetrance and appears usually in adults between the ages of 30-40. Its defect is a repeated sequence of CAG (36 or more times) on chromosome 4 specifically on the Huntington gene. Longer repeat sequence results in an increased likelihood of an earlier presentation of the disease. Symptoms include choreiform movements, mood disorders such as depression and anxiety, aggressive and agitated behavioral changes, cognitive deterioration, ataxia, eye-movement disorders, dysarthria, dysphagia, bradykinesia and dystonia<sup>2</sup>. Treatment is supportive towards the symptoms and there is no cure. Our case was undiagnosed because the symptoms were attributed to a previous stroke.

The aim of this report is to present a case of worsening choreiform movements, imbalance, slurred speech, behavioral changes and cognitive decline.

### THE CASE

A fifty-seven-year-old male with a history of type 2 diabetes, hypertension, dyslipidemia and old ischemic stroke presented to the neurology outpatient clinic with complaints of worsening abnormal movements of both arms since ten years, which worsened in the past three years. Ten years ago, he was involved in a road traffic accident. Imaging had been negative for any brain damage; however, since then, choreiform movements were noticed. Three years ago, the patient had a stroke, resulting in right-sided weakness that completely resolved. Following the stroke, the brisk movements worsened and his condition deteriorated with the onset of additional symptoms. His MRI at the time of the stroke revealed an infarct at the left lentiform nucleus and left deep white matter nuclei. He suffered from imbalance and dizziness, slurred speech and unusual behavior, all of which were attributed to his recent

stroke. Due to dizziness and imbalance, he had multiple falls. He had no history of epilepsy. He was prescribed clonazepam 1 mg OD PO with increasing the dose to BD as required to control his choreiform movements and betahistine for his dizziness. The choreiform movements became progressive, but were attributed to a post-stroke syndrome.

His regular home medications included Metformin 500 mg BD PO, ASA 81 mg OD PO, Clopidogrel 75 mg OD PO, Lipitor 20 mg OD PO, Clonazepam 1 mg BD PO and Betahistine 8 mg BD PO. He has no known allergies.

The patient is married with 3 children aged 35, 32 and 20, none of whom have any choreiform movements. The patient's father had similar movements that were noticed in his 60s and remained undiagnosed. The patient quit smoking after the stroke. He never consumed alcohol and had no history of illicit drug use.

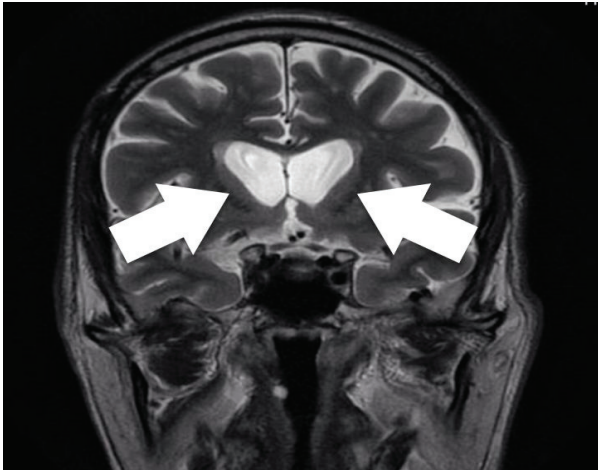
The patient looked nervous, oriented and his memory was preserved. His neurological examination showed normal equal power, tone, and reflexes in all limbs. His sensation was equal and intact in all dermatomes. Cranial nerves were intact. Coordination was preserved and cerebellar signs were absent. Positive findings were the 'dancing' movements of his hands and face, and a sign of Huntington's including fragmentation of saccadic eye movements, milkmaid's hand grip and flycatcher's tongue.

Huntington's disease was suspected especially with the newly discovered family history. MRI brain and genetic CAG trinucleotide repeat test were ordered. The diagnosis was confirmed with the presence of 23 CAG repeats seen in one allele and 41 repeats in the other allele of the Huntington gene.

MRI brain revealed age-related brain involutational changes and chronic white matter low-grade microvascular ischemic changes with a decrease in the size of the caudate nucleus, see figure 1.

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**Figure 1: MRI Brain Showing Atrophy of Caudate in Coronal View**

During subsequent follow-ups, he was still intermittently symptomatic with choreiform movements and had developed some memory impairment as well. His behavioral symptoms progressed and included aggression. He also developed symptoms of anxiety and panic attacks including somatic complaints of tachycardia and shortness of breath with the feeling of impending death. He had thoughts of suicide with other symptoms of depression such as random bouts of crying.

He was prescribed Tetrabenazine 12.5 mg OD with a plan to increase to BD, but this medication was not available in Bahrain. He was also prescribed Quetiapine 25 mg which was increased to 50 mg HS PO. He was referred to psychiatry and family counseling was also done for genetic testing.

## DISCUSSION

Patients who experience a stroke are usually left with various residual symptoms; often due to specific areas in the brain which the ischemia or hemorrhage have irreversibly damaged. In our patient, many of his symptoms were commonly found in post-stroke individuals. These include movement disorders, behavioral and personality changes, altered cognition, mood symptoms and imbalance<sup>3</sup>.

Our patient's symptoms began many years earlier, initially with only the choreiform movements. Additionally, after his stroke, the additional symptoms he experienced did not specifically match the area of his particular stroke, as studies show that lenticular infarcts tend to cause hemiparesis rather than movement disorders<sup>4</sup>. Therefore, other differential diagnoses for his symptoms would have been helpful.

The differential diagnosis in patients with symptoms of choreiform movements needs to be considered in the context of other associated symptoms. Sydenham's chorea was unlikely in our patient as there was no history of Group A Streptococcal infection. Other symptoms including hypotonia, joint pains, and facial grimacing were also absent. Neuroacanthocytosis usually presents with early onset of chorea, and blood tests would reveal normocytic anemia (with a smear showing acanthocytes), neither of which were present in this patient. Hereditary chorea would also have an early onset. Wilson's disease may include findings of jaundice, additional severe

psychiatric symptoms as well as Kayser Fleischer rings. Ataxia telangiectasia would present in childhood with skin lesions and other cerebellar symptoms<sup>5</sup>.

Patient's MRI findings and genetic studies were typical of Huntington's disease. MRI findings in patients with Huntington's typically show atrophy of the caudate and putamen due to loss of GABA-ergic neurons<sup>6</sup>. In our case, MRI and the CAG trinucleotide repeat test confirmed the diagnosis. However, the genetic test is more sensitive and specific in diagnosing this disease.

Huntington's disease has no cure, however, it is important for families to be aware of its implications and decide regarding genetic testing. Family support and awareness of its symptoms would help in anticipating what the patient might experience in the future. Suicide is a common mode of death in Huntington's patients as a result of their depression, and this could be something that families could be made aware to prevent.

Treatment is usually multidisciplinary and supportive for patients as this is a progressive disease. Pharmacological treatment is used to alleviate symptoms and they include dopamine blockers or dopamine-depleting agents such as tetrabenazine, benzodiazepines such as clonazepam, antiepileptic medications and antidepressants. As seen in our patient, symptoms were not always controlled and they need to be followed up and medications adjusted as needed<sup>7</sup>.

## CONCLUSION

**The diagnosis of Huntington's disease was not considered earlier in this case. It is important to acknowledge that although stroke patients very often have many post stroke symptoms, which may not always have a direct designation to a specific brain area; other diagnoses should always be ruled out involving a detailed history.**

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