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Huntington's Disease of Childhood: Awareness and Prevention

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ABSTRACT

Huntington disease is fatal, autosomal-dominant neurodegenerative disorder of adults. During the 1960s, the rare occurrence of childhood and adolescent forms of Huntington disease was increasingly recognized. Pediatric Huntington disease typically has different presentation when compared with adults. Early diagnosis and symptomatic treatment may contribute to enhancing the quality of life of affected patients. However, most clinicians are unaware that Huntington disease can present during childhood and adolescence, and the diagnosis can be delayed. Therefore, appropriate genetic counseling are commonly not witnessed. Pediatric Huntington disease has not been reported before in Iraq. The main aim of this paper is to describe the first case of this disease in Iraq. This case emphasizes the importance of awareness of the disease to help in providing early genetic counseling which may contribute the possible prevention of further cases in the affected family.

KEYWORDS

Pediatric Huntington disease, Iraq, Awareness

INTRODUCTION

Huntington disease is generally an adult onset autosomal dominant genetic degenerative brain disorder presenting with dementia, choreiform movements and gait abnormalities. The disorder is associated with death of brain cells and was also called "Huntington's chorea" because of the associated chorea "dance-like movements" which is uncontrolled movements. However, not all patients develop chorea and cognitive and behavioral symptoms can be the major manifestations.

Although the condition is typically inherited from parents, in about 10% of patients, the condition is caused by an autosomal dominant new mutation causing expansion of CAG (cytosine-adenine-guanine) triplet repeats in the gene coding for the Huntingtin protein leading to abnormal protein. The abnormal Huntingtin protein gradually damages cells in the brain.

The onset of the disease is during the middle age, and symptoms usually appear between thirty and fifty years of age, but may the disorder may appear any age[1,2].

Pediatric Huntington disease has not been reported before in Iraq. The main aim of this paper is to describe the first case of this disease in Iraq.

Case Report

SR was a 17-year old boy with progressive neurological deterioration, first noticed when he was studying at third grade primary school at about the age of nine years. He experienced deterioration in speech and school performance. He had to repeat the third grade, but his speech deteriorated significantly, and he couldn't complete the third grade of primary school and left school.

During the previous years he experienced progressive gait abnormalities, difficulties in feeding and abnormal movements including chorea and athetosis. A difficulty with swallowing was gradually progressed during the previous five years.

During the previous two years, abnormal involuntary movements of the upper and lower limbs became more prominent.

The boy was totally unable to walk alone or say any during the previous four months. Brain CT-scan was performed on the eighth of

February, 2016 was normal. Pure tone audiogram was performed on the first of June, 2016 and showed bilateral normal hearing.

MRI imaging of the cervical spine (Sequences performed: T1, T2 sagittal, and SS myelo) was performed on the 5th of June, 2016 showed hypertrophied adenoids with patent airways.

MRI imaging of the cervical spine without contrast (Sequences performed: T1, T2 sagittal, and SS myelo) was performed on the 25th of October, 2016 showed loss of normal cervical lordosis mostly due to muscle spasm.

Brain CT-scan was performed again on the 27th of October, 2016 showed slightly dilated ventricular system with prominent axial CSF spaces suggesting mild brain atrophy.

MRI imaging of the brain with a diffusion study was performed on the 20th of August, 2017 showed normal brain parenchyma signal intensities with no area of restricted diffusion.

MRI imaging of the brain performed on the 21st of November, 2017 showed:

- Mild diffuse supra-tentorial brain atrophy with no changes in brain signal intensity.
- Mild hypoplasia of the cerebellar vermis with normal cerebellar hemispheres.

Brain CT-scan was performed again on the twelfth of August, 2018 showed mild, but diffuse brain atrophy. When he was first seen on the 25th of October, 2018 Figure-1), he was unable to walk, unable to eat, and unable to say a single word. He had rigidity and his mouth was kept open most of the time, and was having choreoathetoid movements and extensor spasm of the neck.

The child was passively receiving fluids orally by his mother. The parents were relatives. The patient was one of two male twins, the second twin died before about ten years with his father by an explosion. The father died early in his thirties and he didn't show obvious neurological deterioration.

He also had a normal 16-year old sister. Other siblings included a twin boy and girl who were studying at third grade primary school and have recently developed speech abnormality mainly in the form of stuttering in association with some deterioration in school performance. The mother was not having any obvious illness.

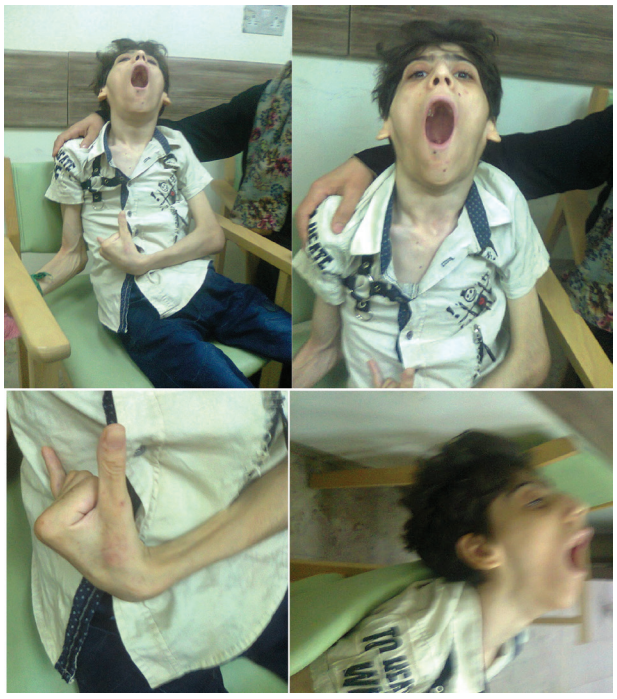

Figure 1: The patient was unable to walk, unable to eat, unable to say a single word, and his mouth was kept open most of the time. He had had choreoathetoid movements, and was developing extensor spasm of the neck

DISCUSSION

The familial disorder of involuntary movements and dementia affecting only adults was named George Huntington. However, a juvenile or adolescent form was thought initially by some to be a separate clinical entity, but has also been associated with the name Huntington disease despite attempts to re-classify the disorder. During the 1960s many papers reporting the occurrence of childhood and adolescent forms of Huntington disease were published in English and other languages including French (Brion and Comoy, 1965),German (Schönfelder, 1966; Lehmann, 1966;Gröpl, 1968), Italian (Saginario,1967; De Marco and Testa, 1969),Serbian (Rodojicić and Bumdasirević,1969) [2].

Barrows and Cooper (1963) called the pediatric disorder appropriately Huntington's disease of childhood avoiding the misleading word "chorea" as rigidity can be more prominent in the pediatric disorder [3].

Children with pediatric Huntigton disease initially develop normally, but becomes clumsy, ataxic, develop dysarthria, and mental retardation [2].

Quigley J (2017) from the University of Michigan defined juvenile Huntington disease as a neurodegenerative disease with onset before the age of twenty one years, therefore it includes the pediatric cases. The juvenile disease accounts for a minority of Huntington disease diagnoses. Juvenile Huntington disease typically has different presentation when compared with adults. It is characterized by cognitive and behavioral changes, parkinsonism, oropharyngeal dysfunction, and seizures [4].

The distinctive clinical features of Juvenile Huntington disease include [2,4]:

Juvenile generally progresses more rapidly and chorea may appear for short and rigidity can be the dominant symptom. Rigidity, dystonia, and seizures are commoner in the juvenile form of Huntington disease. Cognitive impairments and deterioration in school performance are common.

Early diagnosis and symptomatic treatment may contribute to enhancing the quality of life of affected patients. However, most clinicians are unaware that Huntington disease can present in childhood and adolescence, and the diagnosis can be delayed. Therefore, appropriate genetic counseling are commonly not witnessed.

This case emphasizes the importance of awareness of the disease to help in providing early genetic counseling which may contribute the possible prevention of further cases in the affected family.

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