

Juvenile-Onset Huntington's Disease in a 12-Year-Old Girl: A Rare Case Report

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ABSTRACT

Huntington Disease (HD) is an autosomal dominant neurodegenerative disorder caused by CAG repeat expansion in the HTT gene. Juvenile Huntington disease (JHD), defined by onset before 20 years, is uncommon and often differs from adult-onset disease, with cognitive, behavioural, rigidity, ataxia, and epileptic manifestations predominating over chorea. We report a 12-year-old girl presenting with a three-month history of mild bilateral action tremor and a two-month history of declining academic performance, accompanied by irritability and social withdrawal. Neurological examination revealed mild choreiform finger movements, axial hypotonia, subtle saccadic slowing, impaired tandem gait, and mild dysarthria. The Unified Huntington's Disease Rating Scale motor score was 18, while cognitive screening demonstrated deficits in attention and executive function. Genetic testing confirmed a pathogenic HTT CAG expansion of 62 repeats, with 18 repeats on the normal allele, establishing JHD. There was no known family history of HD. Cascade testing of the clinically asymptomatic 54-year-old mother unexpectedly identified a 39-CAG repeat allele within the reduced-penetrance range. The marked intergenerational expansion from 39 to 62 repeats and absence of an apparent family history highlight genetic anticipation and clinically silent parental carriage. Although paternal transmission predominates in JHD, this case demonstrates that substantial maternal transmission can occur. It also highlights the diagnostic importance of recognizing academic and behavioural deterioration as early manifestations of JHD and the role of cascade testing and genetic counselling in apparently sporadic pediatric cases.

INTRODUCTION

Huntington Disease (HD) is an autosomal dominant neurodegenerative disorder caused by pathological CAG repeat expansion that encodes the HTT gene [1]. Juvenile Huntington's disease, defined as HD with an onset of ≤ 20 years of age, is a relatively rare condition characterized by features such as epilepsy, ataxia, spasticity, and other atypical motor and behavioural manifestations [2]. Disease progression is faster in JHD as compared to adult-onset HD, and cognitive decline is detected primarily by declining school performance, which was consistent with our case as well [3]. Studies conducted in the past by Fusilli et al. [4] and Went et al. [5] state that around 80% of JHD patients inherit the repeat expansion via paternal transmission [4,5].

In contrast, our case describes a 12-year-old girl with a 62 CAG repeat expansion and no known family history, whose clinically asymptomatic mother was subsequently found to carry an intermediate-to-expanded, 39 CAG repeat allele on genetic testing. This unusual maternal transmission highlights the phenomenon of intergenerational CAG repeat expansion and underscores the importance of genetic counseling and cascade testing in cases of sporadic JHD.

CASE DESCRIPTION

We report a 12-year-old female who presented with a three-month history of mild bilateral hand tremors and a two-month history of declining academic performance, noted by teachers as difficulty with concentration, slowed processing, and deterioration in handwriting. Parents also reported subtle behavioral changes, including increased irritability and social withdrawal over the preceding two months. The tremors were fine, action-related, and not disabling, with no impact on activities of daily living. There were no history of seizures, vision changes, or bowel and bladder involvement.

Neurological examination revealed mild choreiform movements of the fingers and mild axial hypotonia, with subtle oculomotor slowing on saccadic testing; gait was mildly unsteady on tandem walking but otherwise preserved. Speech was fluent with mild dysarthria on rapid repetition tasks. Formal assessment using the Unified Huntington's Disease Rating Scale (UHDRS) yielded a total motor score of 18, consistent with early symptomatic disease, and cognitive screening (MoCA) showed mild impairment in attention and executive function, correlating with the reported academic decline. Given the combination of motor, cognitive, and behavioral features, Juvenile Huntington's Disease (JHD) was suspected, and genetic testing was performed.

Genetic analysis confirmed a pathogenic CAG repeat expansion in the HTT gene, with 62 repeats on the expanded allele and 18 repeats on the normal allele, confirming a diagnosis of juvenile-onset HD. There was no prior family history of HD reported at presentation, and the child's mother, aged 54 years, was clinically asymptomatic with no motor, cognitive, or behavioral complaints. In view of the child's genetically confirmed diagnosis, cascade genetic testing was offered to the mother for counselling purposes.

Unexpectedly, the mother was found to carry an intermediate-to-expanded CAG repeat allele (39 repeats) on genetic testing, an incidental finding in the absence of any prior symptoms or known family history of HD. This finding raised the possibility of a de novo or unrecognized paternal/maternal lineage expansion and prompted further genetic counselling regarding anticipation, given the marked difference in repeat length and age of onset between mother and child. The family was referred for multidisciplinary follow-up, including neurology, genetics, and psychosocial support.

DISCUSSION

Juvenile Huntington's Disease (JHD) accounts for roughly 5% to 10% of all HD cases and is defined by symptom onset before the age of 20 [1]. Compared with adult-onset disease, JHD more often presents with rigidity, seizures, and cognitive or behavioural change rather than florid chorea [1]. This pattern was reflected in the present case, where academic decline and irritability preceded any noticeable motor abnormality, and the choreiform movements found on examination remained mild throughout. The attentional and executive deficits on MoCA are also consistent with what has been described in JHD, where decline in school performance is often the first clue to cognitive involvement [1].

A CAG expansion of 62 repeats sits well within the range associated with juvenile onset, and repeat length correlates inversely with age at onset - larger expansions generally produce earlier and more severe disease [6]. What makes this case somewhat unusual is that the expanded allele was maternal in origin. Most JHD cases arise from paternal transmission, largely because the CAG repeat is more prone to further expansion during spermatogenesis than oogenesis [6]. Large maternal expansions leading to JHD do occur, but they are distinctly uncommon, which is part of why they tend to get reported individually [7].

The mother's own result is arguably the more interesting finding here. Her 39-repeat allele falls in the reduced-penetrance range, where a carrier may remain asymptomatic for life or go on to develop only a mild, late-onset illness [8,9]. At 54, with no motor, cognitive, or behavioural complaints, she fits that pattern well. It also explains the absence of a known family history at presentation - a person can carry a reduced-penetrance allele for decades without anyone in the family suspecting HD.

This case is a reminder that a negative family history does not exclude HD, since a parent may silently carry a reduced-penetrance allele capable of expanding further on transmission. It also illustrates the value of cascade testing after a pediatric diagnosis, which can uncover carriers who would otherwise never have come forward for testing, and who then need genetic counselling of their own regarding recurrence risk and anticipation. Ongoing multidisciplinary follow-up involving neurology, genetics, and psychosocial support is warranted for both the child and the wider family.

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