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Case Report

A Case Report on Huntington's Disease Presenting with Progressive Motor and Cognitive Decline

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Background of study

An uncommon, hereditary, autosomal dominant neurological condition, Huntington's disease (HD) is marked by increasing motor dysfunction, cognitive loss, and mental symptoms (Stoker et al., 2022). A trinucleotide (CAG) repeat expansion in the HTT gene on chromosome 4 is the cause of the illness (Kumar et al., 2020). Clinical onset usually happens in mid-adulthood, and symptoms steadily get worse over time, eventually resulting in death and severe disability (Ogilvie et al., 2023). One important diagnostic characteristic is a rich family history spanning several generations (Kringlen et al., 2017). Genetic counseling, family screening, and proper symptom treatment all depend on early detection of HD.

Case Presentation

A 47-year-old man with a history of increasing involuntary movements and cognitive deterioration arrived at the Neurology Outpatient Department of Lady Reading Hospital (LRH), Peshawar. The patient was a resident of Ghazi Dheri, Tehsil Tangi. The patient claims that his symptoms began around ten years ago and initially manifested as involuntary movements before extending across his entire body. Slurred speech accompanied these motions, which become more noticeable with time. Cognitive involvement was also indicated by the patient's complaints of difficulties focusing, poor work planning and organization, memory issues and forgetfulness. The patient also had back ache. A strong positive family history of a comparable neurological condition was identified in the patient. He has two sons and two daughters. He had two sisters and six brothers, just one of them is still alive. Particularly, the patient's mother, four of his brothers and maternal aunt all experienced identical symptoms and have since passed away. A genetic pattern is further supported by the fact that two nieces are now affected. The family has a history of consanguineous marriages. The patient displayed broad choreiform movements, dysarthric speech and cognitive impairment, specifically affecting executive functions and memory, throughout the neurological examination. The patient's clinical appearance, increasing course, and noteworthy family history led to the diagnosis of Huntington's disease.

Discussion

Huntington's disease is a neurological disorder with characteristic behavioral, cognitive, and mote symptoms that worsens with time (Walker et al., 2007). Chorea is often the most prevalent early motor indication, followed by speech issues and cognitive impairment, particularly with relation to executive functioning and memory (Roos et al., 2010). The patient's age of onset, gradual deterioration over a ten-year period, and cognitive decline are consistent with Huntington's disease. The significant intergenerational engagement in this case is a noteworthy feature that strongly supports an autosomal dominant inheritance pattern. Consanguinity may have contributed to the family's increased

sickness burden. Cognitive symptoms include poor attention, difficulty forming plans, and forgetfulness have a significant influence on functional independence and quality of life. This case emphasizes how important it is to recognize classic clinical indications and family history when diagnosing Huntington's disease, particularly in low-resource areas where genetic testing may not be readily available.

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