

# A Rare Case of Huntington's Disease Associated with Depression and Psychotic Features

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## ABSTRACT

Huntington Disease is rare chronic neurodegenerative disease, characterized by triad of abnormal choreatic movements, cognitive and psychotic disorders. Noteworthy, in spite of typical motor symptoms other than chorea have been reported, a very small group presents with both depressive and psychotic features. In this paper, we present a 46 Year old female patient of confirmed diagnosis of HD after molecular evidence of CAG report comprising depression and psychotic attributes.

**Key Words** – Huntington diseases, depression, psychosis

## INTRODUCTION

Huntington disease (HD) was first described by George Huntington in 1872 as a neuro-degenerative disorder comprised by triad of unwanted choreatic movements, cognitive and psychiatric characteristics. The disease is caused by increased Cytosine Adenosine Guanine (CAG) trinucleotide in gene IT15, which encodes a protein 'Huntington' having an unknown function. [1] The main pathology is the neuronal loss occurs from the striatum, particularly in caudate nucleus leading to neuropathological changes in cortical and subcortical regions causing cognitive and movement impairment. Its prevalence in the different demographics of the world ranges from 5 to 10 per 10,000. [2,3] HD can prevail at any age, but usual age of onset is midlife i.e. 30- 40 Yrs of age group. [4] The symptom progresses to complete disability and death typically in 15-20 Years. Present treatment options are found to be insufficient to inhibit the progression of disease. Hereby, in this article we present a case of Huntington disease masquerading with depression and with psychotic features.

## CASE REPORT

A 46 year old female, married, illiterate living in nuclear family in rural area of Rajasthan, India presented with complains of abnormal dance like movement which was non- repetitive, non – periodic associated with occasional fall from 11 years. These abnormal movements had initially begun from the left hand and progressed to both hands.

Over the time the movements sequentially involved upper limbs , lower limbs , neck and face and was finally seen in lips and tongue with tremor.

During the initial stage, patient reported with depressive symptoms like sleep disturbances, low mood, loss of interest, irritability and fatigue.

As the disease progressed patient began to have psychotic features like Suspiciousness, aggressiveness, poor self-care, remains aloof, hearing of voices which are persecutory in nature.

Patient had a positive family history of abnormal movements in father and grandfather and died at the age of 62 and 80 respectively.

Mental status examination – showed unclear speech, increased psycho- motor activity, blunted affect, delusion of reference and persecution. She had auditory hallucination 3<sup>rd</sup> person, Impaired cognition and insight was absent.

On Examination:-CNS–High motor function-Normal, Speech-hypo-phonic, decreased word output, speaks in short sentences. Cranial nerve-pupil B/L Equal and reactive towards light, No ptosis, No nystagmus, Tone-hypotonic, Power -5/5 in all limbs, Deep tendon reflex-decrease, Plantar -Right and left flexor, Sensory-normal for pain, touch temperature, vibration, joint point sensation .Cerebellum –Normal skull and spine, no neck rigidity and Kernig's sign, Gait- ataxic. Involuntary movement-abnormal non-repetitive, non-periodic, large amplitude dance like movement in all limbs (chorea). CVS – S1,S2 +, No murmur. Respiratory system – B/L Air Entry +,No Crepitations. Fundus examination – Normal.

Investigation – MRI (magnetic resonance imaging) of brain revealed caudate atrophy with ballooning of frontal horn of lateral ventricles. Chest X-ray and 2-D Echo was normal. Ultrasonography for abdomen was normal, Thyroid Profile (T3, T4, and TSH) was normal and other blood tests including CBC, RBS, RFT and LFT were found to be normal.

Diagnosis of HD was confirmed by PCR (CAG) trinucleotide molecular analysis which showed 21 CAG repeats in one allele and 46 allele of Huntington gene. Considering the family & patient history, clinical examination, MRI findings and CAG report, diagnosis of Huntington chorea was made.

Patient was managed with Tab. Risperidone (upto 8mg), Tab. Sertraline (upto 200 mg), Tab clonazepam (upto 4mg), Tab. Tetrabenazine-(50 mg) in divided doses. Patient had shown significant symptomatic relief in the psychotic and

depressive features and now she is able to carry out household chores.

## DISCUSSION

Huntington chorea is a autosomal dominant disorder predominant with motor symptom, positive family history and molecular genetic analysis.

According to Aggrawal et.al 2004, [5] Cause of hereditary chorea includes:-

The common cause of hereditary chorea includes

- Neuroacanthocytosis
- Benign hereditary chorea
- Huntington's like disease and

For Rare causes are –

- Fahr disease
- Spinocerebellular ataxia
- Wilson Disease
- Neuronal ceroid lipofuscinoses
- Ataxia telangiectasia
- Haller vordenspatz disease
- Dentatorubral-Pallidoluysian atrophy(DRPLA)

Benign hereditary chorea is characterized by onset in childhood. In ataxia telangiectasia patient presents with continuous telangiectatic lesions with degenerative cerebellar function and immunodeficiency. Fahr disease is idiopathic basal ganglia calcification whereas Wilson disease autosomal recessive pattern with neurological and psychiatric symptoms. [6,7]

The description of this case illustrates the broad range of clinical presentation that satisfies need for considering HD as a diagnosis of patient. Patient has substantiated family history (Fig: 1) and Huntington gene with CAG repeats varying in size. In general population, there is a mean of around 19 CAG repeats with average of 9-37. In HD average length is 46 and range up to 36-86. [7]

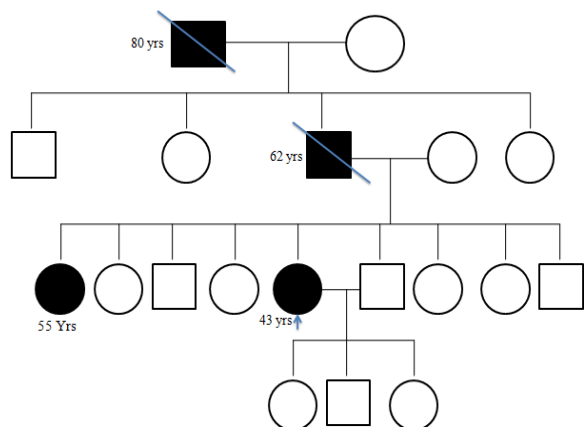


Fig: 1. Family Tree

According to Negi et.al 2012 [8] Imaging Studies remains cornerstone in diagnosis and severity of Huntington chorea. Case reveals atrophy of caudate from frontal horn to lateral ventricles is seen (Fig: 2a and 2b).

Abnormal movements which are first and most evident in hand and face. [9] Accompanied with disturbance of mood, comprising depression. [10] Irritability and aggression. [11]



Figure :2A

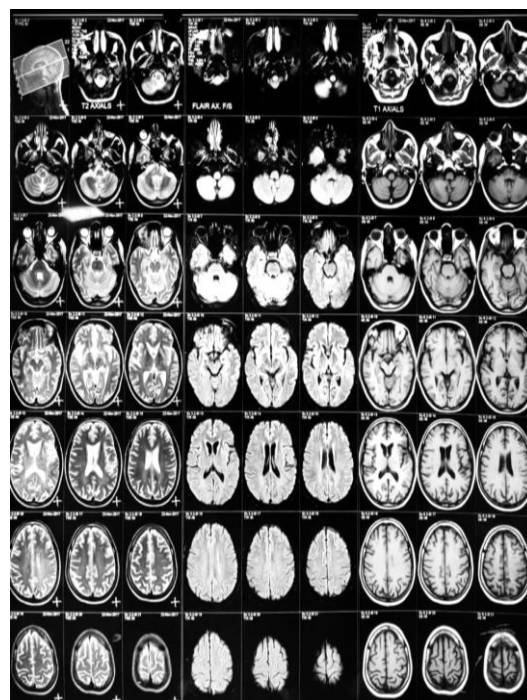


Figure – 2B

The diagnosis was made clinically and confirmed by MRI and CAG report, CAG expansion has a sensitivity of 98.8% as a diagnostic test and it is highly specific. Huntington disease is progress steadily with other co morbidity. The Patient should be provided with psychological and supportive treatment for managing associated debilities, Genetic counseling is important for better outcome.

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