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# Huntington Disease and Genetical Relation- A Case Study in Kerala

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**ABSTRACT**
The meticulous therapy for Huntington Disease is still beyond the knowledge. Huntington disease is a neurodegenerative disorder characterized by choreatic movements, behavioural and psychiatric disturbance. The present case was observed on 34 year old women, from a family in which the three previous generations were affected by the same disease.She showed the symptoms at the age of 26 and was diagnosed by means of signs and family history. All other organs tests were unremarkable. Symptomatic therapy is the only treatment existing at present thus she was discharged with tablet Lonazep (Clonazepam) 0.5mg .Counseling to the family plays a chief role since there is 50-50 chances of getting this disease to the next generations.
**KEYWORDS:**Huntington disease, choreatic

## I. INTRODUCTION

Huntington disease is an incurable disease which occurs due to the degeneration of nerves in the brain. Huntington disease is caused by the mutation of HTT gene. This HTT gene makes proper information for the production of a protein called huntingtin[3]. Expansion of CAG segment results in an abnormal protein formation which leads to brain cells damage. In normal conditions , the CAG segment is repeated 10 to 35 times within the gene but in patients with Huntington disease, it will repeated 36 to more than 120 times. Patients with 36 to 39 CAG repeats may or may not progress the signs and symptoms, while people with 40 or more repeats develop the disorder . In the present study in Kerala, a rare case of juvenile form of Huntington disease had identified and having CAG segment repeats of 51 and case report had prepared with the slight intervention of study of family history also[2]

## II. METHODS

### a. Case study

Case was reported in a tertiary care hospital few years back.A 34 years old women presented to outpatient department over the complaints of abnormal jerky movement of both upper and lower limbs, can't able to hold things, rapid eye movement, laterocollis, labial dyskinesia. She had history of Huntington disease for the past eight years.

Family history showed that she acquired through paternal link in family tree. Her grandfather died at the age of 38 and her father died at the age of 30 and her younger brother died at the age of 22, and her daughter died at the age of 12, all of them were reported to have same symptomaticdisease as depicted in figure 1.

### b. Diagnosis of Huntington Disease

Diagnosis of Huntington disease is based on the clinical sign and symptoms, family history, scan reports, Huntington gene test[1].

On examination MMSE ( Mini – Mental State Examination) scale shows 29/30 which shows no dementia , UHDRS ( Unified Huntington Disease Rating Scale) also done , HD gene test positive , number of CAG repeats 51,spinocerebellar ataxia (2,3,17,20) . Her CVS, respiratory system were regular. Chest X ray and ECG showed no cardiac deviation. UltraSound Sonography for abdomen was normal.

Patient stayed on regular follow up for the past 8 years and is being treated with Lonazep 0.5mg three times a day

**1. Pedigree study of the patient's family**

**Inheritance Pattern**

Huntington Disease is inherited by an autosomal dominant manner, that is having a mutation in only one of the two copies of the HTT gene is enough to cause HD. In the present case, the number of CAG repeats of the patient was reported to be 51 .

If a person with HD has children, each child has a 50% chance to inherit the mutated gene and develop the condition[2], here her brother also affected with the same disease.

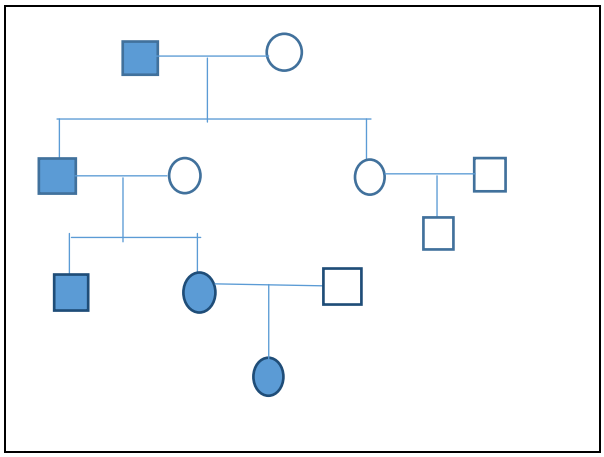

Fig 1: The family tree of the reported patient. (Affected one : filled with blue colour, Square box : male, Circle : female)

## III. DISCUSSION

Huntington disease was named after George Huntington in 1972 .For the first time, actual diagnosis made and the disease involving trinucleotide repeats of CAG were found. C (cytosine ), A ( adenine ), G ( guanine ) is the trinucleotide[3].Huntington disease is accompanying with 36 repeats or more. In thispatient the number of CAG repeats is 51. Surgical treatment does not play an important role in Huntington Disease.

This disorder is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is adequate to cause the disorder. An affected person typically inherits the altered gene from one of the affected parent.As the altered HTTgene is delivered from one generation to the next, the size of the CAG trinucleotide repeat often increases[4] . A higher number of repeats leads to an earlier onset of signs and symptoms.

Cases of early onset are in most occurrences transmitted by the father.In this case also it is transferred from patient's father. Paternally transmitted cases have a more number of CAG repeats[5] .Our patient, who inherited the disease from her father, is therefore expected to progress more speedy, is called as anticipation that is the tendency for autosomal dominant diseases to be graduallymore severe in following generations[6] .

Stages of Huntington Disease

| | | |
|---|---|---|
| Stage 1 | Early stage | Onset of disease, fully functional at home and work |
| Stage 2 | early intermediate stage | Last between 3-13 years, patient still works normally |
| Stage 3 | late intermediate stage | Last between 5-16 years, patient no longer can work properly. |
| Stage 4 | early advanced stage | Last between 9-21 years , patient require additional support |
| Stage 5 | Advanced stage | Last between 11-26 years, patient need total support to perform daily activities. |

Patient with Huntington Disease usually die within 20 years due to the occurrence of complications such as infection, heart failure, pneumonia.

Now this patient is on stage 5 that is on advanced stage as she is not able to walk or perform activity by herself.

## IV. CONCLUSION

The present case was observed on a women from a family in which the three previous generations were affected by the disease as well showing the importance of family counseling. Our patient′s family was not aware about the genetic relation of this disease .Vertical transmission of Huntington disease is understood through three generations of this patient's family. If the ages ofonset of Huntington disease is at young and if this disease is diagnosed at the early stages , at least the patient can be treated with psychological and other supportive measures . Genetic counseling of course would go a long way in the eradication of this devastating disease.

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