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HUNTINGTON DISEASE –CASE REPORT AND EVALUATION OF MRI BRAIN PARAMETERS

Neurology

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ABSTRACT

We report a genetically confirmed case of Huntington's disease and its evaluation of MRI brain parameters. Huntington's disease (HD) is an autosomal dominant disorder characterised by involuntary movements, dementia, and behavioural disturbances. MRI Brain parameters (FH/CC AND CC/IT RATIOS) are very specific for evaluation of HD and adds to the diagnosis OF HD. Diagnosis can be confirmed by genetic testing of CAG nucleotide repeats.

KEYWORDS

Huntington disease, Autosomal dominant, CAG nucleotide repeats, MRI Brain parameters.

CASE REPORT-

- A 26 years old young male patient presented with slurring of speech, intermittent unsteadiness while walking, intermittent fidgety movements of extremities, behavioural issues in the form of irritability, excessive anger, mood liability and chronic drug abuse since 2 years.
- There was no history of fever /vomiting/ seizures, bladder, and bowel incontinence.
- Family history was positive in three generations that is in grandfather and father. However they were not evaluated.
- Neurological examination reveals slurring of speech, and ataxia of gait and limbs.

Patient then underwent MRI Brain which revealed-

- Bilateral caudate head atrophy resulting in enlargement of the frontal horns, giving “box" like configuration (Figure 1).

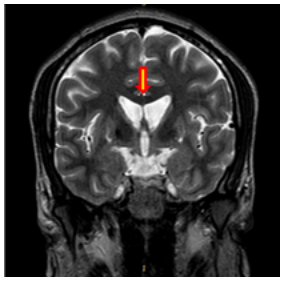

Figure 1

- This was quantified by MRI parameters(Figure 2):
  Frontal horn width to intercaudate distance ratio (FH/CC) which was 1.6
  Intercaudate distance to inner table width ratio (CC/IT) which was 0.19

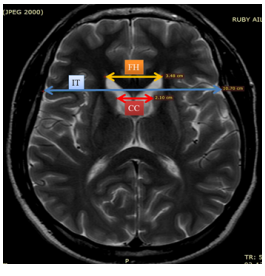

Figure 2

On Huntington disease mutation analysis, patient was found to have excessive CAG repeats with full penetrance which confirmed the diagnosis of Huntington disease.

DISCUSSION:-

Huntington disease (HD) is an autosomal dominant disorder which is characterised by involuntary movements, dementia, and behavioural disturbances. It is caused by a mutation in IT15 gene, which leads to unstable CAG trinucleotide repeat expansion.

Huntington's disease is suspected in a person who presents with cognition changes, chorea and behavioural or psychiatric problems such as depression, irritability, or mood swings, and when there is a positive family history of HD in a parent. MRI brain shows specific changes in caudate nucleus and putamen [1]. Other findings which can be seen are reduction of hippocampal, entorhinal cortex; cerebellum and brainstem volume [2, 3, 4].MRI Brain parameters are very specific for evaluation of HD and add to the diagnosis.

MRI Brain parameters are:-

- Ratio of frontal horn width (FH)/intercaudate distance (CC) - The normal mean FH/CC ratio range is 2.2 to 2.6. As the caudate heads reduce in volume the CC distance will approach the FH distance, and the ratio will approach 1(decrease).
- Ratio of intercaudate distance (CC)/inner table width (IT) - The normal CC/IT ratio range 0.09 to 0.12. As the size of caudate heads reduces, the CC distance will increase and as such the CC/IT ratio will increase.

In our case FH/CC ratio IS 1.6 (Decreased) and CC/IT ratio IS 0.19(Increased) which suggests the diagnosis of HD.

Diagnosis can be confirmed by genetic testing of CAG nucleotide repeats [5]. The number of CAG repeats in normal alleles can range from 10 to 26. CAG repeats between 27-35 in a patient increases the risk of developing HD in their offspring. However, more than 60 CAG repeats in a symptomatic person confirm the diagnosis. As in our case number of CAG repeats was 75. Age of onset of disease symptoms is usually inversely proportional to number of CAG repeats. [6].

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