Huntington Disease : A case report.

CLINICAL HISTORY: A 32 year old female patient, presented with history of abnormal involuntary stereotyped movements, memory loss and emotional instability.

Fig1 : T1W IR Coronal , Fig 2: T2Wi Axial . Fig 3 : ADC Axial: Loss of volume of head of both caudate nuclei and putamina is seen. Increased ADC values are seen in both caudate nuclei in Fig 3.

PLAIN MRI FINDINGS:
Loss of bulk of head of both caudate nuclei and putamina was seen.
Increased ADC values were seen in the head of caudate nuclei.
The frontal horn appeared prominent which showed box like configuration.
The frontal horn width to intercaudate distance ratio was decreased to 1.5.
Increased intercaudate distance to inner table width ratio to 0.18.
Rest of the brain parenchyma was normal.

FINAL DIAGNOSIS: Huntington Disease, named after George Huntington: American physician (1850 - 1916).

DISCUSSION:
Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disease. It is caused by a loss of GABAergic neurons of the basal ganglia, especially of the caudate nucleus and putamen.
**Epidemiology and Pathology:** Huntington disease has a worldwide prevalence of 5 - 10 per 100,000, and is typically diagnosed between 30 and 50 years of age. Incidence is equal in both genders. The mutation that causes the disease was identified on Chromosome 4p16:3, and consists of a CAG trinucleotide repeat. The juvenile form of the disease is also known with onset at or before age 19 years, in approximately 5% of all cases. In juvenile cases having inherited the disease from the father is far more common, hence it raises the possibility of additional X-linked recessive modifying gene in its pathogenesis.

**Clinical presentation:** Presentation is typically with progressive rigidity, choreoathetosis (Huntington's chorea), dementia, psychosis and emotional instability. The juvenile form has a different presentation, with cerebellar symptoms, rigidity and hypokinesia being prominent.

**Radiographic features:** The heads of caudate are atrophied with enlargement of the frontal horns (often giving them a "box" like configuration), along with a more generalized cortical atrophy. A decrease in the frontal horn width to intercaudate distance ratio (FH/CC), or increase intercaudate distance to inner table width ratio (CC/IT) may also be used, although this is not frequently used in everyday clinical practice.

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.

The normal CC/IT ratio range 0.09 to 0.12. As the caudate heads reduce in size, the CC distance will increase and as such the CC/IT ratio will increase.

MR Spectroscopy: may demonstrate elevation of lactate in the occipital cortex, which correlates with duration of symptoms.

**Treatment and prognosis:** The adult onset form is slower in its course and inevitably leads to death in 14 - 15 years, whereas the juvenile form has a more rapidly progressive course, with death occurring in 7 - 8 years.

**Differential diagnoses:**
- Wilson disease: more commonly involves white matter, thalamus, cerebellum and brainstem.
- Leigh disease: more commonly involves white matter, thalamus, cerebellum and brainstem.
- Hallervorden-Spatz syndrome
- Acute hypoxic encephalopathy
- Carbon monoxide poisoning
- Hypoglycaemic encephalopathy

Regards,

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N.B: This case is authentic and from the archives of Radiance Diagnostics. For any queries/suggestions / feedback write to us at radiance@radiancediagnostics.in. Case of the month can also be accessed anytime online at VIEW BOX at www.radiancediagnostics.in