



Huntington Disease

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Why in News

A team of scientists from the **National Centre for Cell Science (NCCS), Pune** studied the HTT gene in **fruit flies**.

Mutations in the HTT gene (also called Huntington or HD gene) cause Huntington Disease (HD).

Huntington Disease

- Huntington Disease (HD) is a **progressive genetic disorder** which affects the **brain**.
- It causes uncontrolled movements, impaired coordination of balance and movement, a decline in cognitive abilities, difficulty in concentrating and memory lapses, mood swings and personality changes.
- The **HTT genes** are involved in the **production of a protein called huntingtin**. They provide the instruction for making the protein.
 - Mutated genes provide faulty instructions leading to production of abnormal huntingtin proteins and formation of clumps.
 - These clumps disrupt the normal functioning of the brain cells, which eventually leads to death of neurons in the brain, resulting in Huntington disease.
- **No cure exists**, but drugs, physiotherapy and talk therapy can help manage some symptoms.

Key Findings

- In the study on fruit flies, it was observed that the pathogenic Huntingtin protein causes a decrease in the overall protein production in cells.
- The Huntingtin clumps collect together (sequester) molecules of another protein called **Orb2**, which is also involved in the process of protein formation.
 - Orb2 protein is **crucial for maintenance of memory in fruit flies**.

- In humans, a **family of proteins called CPEB** is equivalent to the Orb2 protein in fruit flies.
Further studies found that the CPEB proteins are also sequestered by the pathogenic Huntingtin clumps, similar to the Orb2 protein molecules.
- The study, thus, becomes relevant to and valuable in understanding HD in humans.

National Centre for Cell Science

- It is a national level, biotechnology, tissue engineering and tissue banking research center located at **Savitribai Phule Pune University, Pune.**
- It is one of the premier research centers in India, which works on cell-culture, cell-repository, immunology, chromatin-remodelling.

Source: PIB