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A KEY CELLULAR MECHANISM IN HUNTINGTON DISEASE UNRAVELLED

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Ministry of Science & Technology

A key cellular mechanism in Huntington Disease unravelled

Huntington disease (HD) is a progressive genetic disorder affecting the brain

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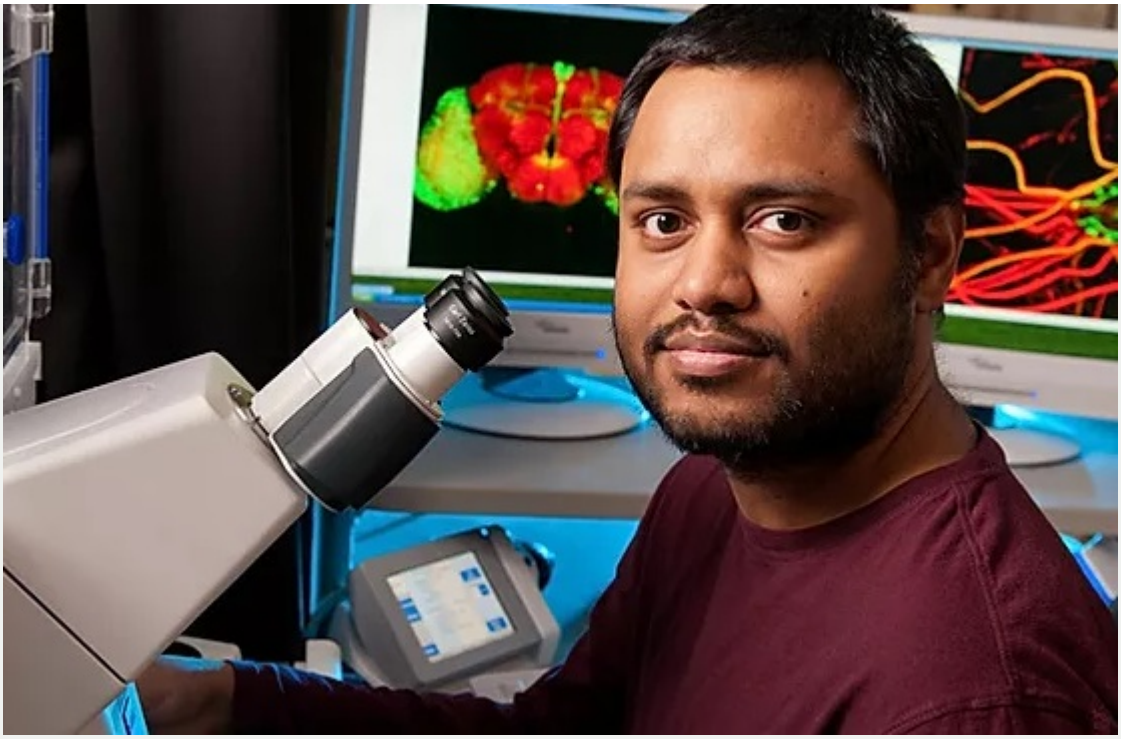
Huntington disease (HD) is a progressive genetic disorder affecting the brain that causes uncontrolled movements, impaired coordination of balance and movement, a decline in cognitive abilities, difficulty in concentrating and memory lapses, mood swings and personality changes.

It is caused by a mutation in a gene called HTT. The HTT genes are involved in the production of a protein called huntingtin. They provide the instruction for making the protein. When the genes mutate, they provide faulty instructions leading to production of abnormal huntingtin proteins and these form into clumps. The clumps disrupt the normal functioning of the brain cells, which eventually leads to death of neurons in the brain, resulting in Huntington disease.

While it is known that the clumps formed by the abnormal huntingtin protein disrupt several cellular processes, it is not known whether they also influence the key process in the formation of other proteins in the cell.

A team of scientists from National Centre for Cell Science (NCCS) in Pune led by Dr. Amitabha Majumdar have been working to gain insights into this by studying the HTT gene in fruit flies. They observed that the pathogenic Huntingtin protein causes a decrease in the overall protein production in cells and that the Huntingtin clumps collect together (sequester) molecules of another protein called Orb2, which is involved in the process of protein formation.



They speculated that the Huntingtin clumps were possibly making molecules of Orb2 unavailable to carry out their normal function associated with protein formation, leading to the observed reduction in proteins in the cell. To gain clarity on this, they induced the cells to produce Orb2 in excess, and found that this did indeed reduce the adverse effects of the faulty Huntingtin protein, which supported their speculation.

In humans, a family of proteins called CPEB is equivalent to the Orb2 protein in fruit flies. The scientists conducted further studies and found that CPEB proteins are also sequestered by the pathogenic Huntingtin clumps, similar to the Orb2 protein molecules. This suggests that the insights gained through the studies carried out by this group in fruit flies are relevant to and valuable in understanding HD in humans.

lii. Orb2 protein is also crucial for maintenance of memory in fruit flies. Therefore, sequestration of Orb2 by Huntingtin clumps may hold relevance to the memory-related issues associated with HD as well. The findings of Dr. Majumdar are expected to pave the way for further exploration to understand this disease better.

The research team has published a report on their work in Cellular and Molecular Life Sciences The team included Hiranmay Joag, Vighnesh Ghatpande, Meghal Desai, Maitheli Sarkar, Anshu Raina from NCCS and Mrunalini Shinde, Ruta Chitale, Ankita Deo and Tania Bose from S.P.Pune University.

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