

New insight into the cellular defects in Huntington's disease

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Huntington disease is a devastating neurodegenerative disorder that causes a progressive loss of functional capacity and reduced life span. It is an inherited condition caused by a mutant HTT gene. Although this has been known for many years, the functions of the normal Htt protein and the mechanisms by which the mutant protein generated from the mutant HTT gene causes disease are not well understood. A team of researchers led by Frédéric Saudou, at the Institut Curie, France, has now uncovered a new function for normal Htt protein and determined that this function is disrupted in a mouse model of Huntington disease and in patients with the disorder.

Detailed analysis by Saudou and colleagues determined that normal Htt protein regulates the formation of cellular structures known as cilia and that cilia were longer and disorganized in the mouse model of [Huntington disease](#) and patients. They therefore suggest that abnormal cilia could be a cause of some of the symptoms of Huntington disease. However, they also caution that further studies are needed to prove this. This point is also made in an accompanying commentary by Scott Zeitlin and Jeh-Ping Liu, at the University of Virginia, Charlottesville, who go on to note that determining this is critical to discerning whether therapeutic strategies designed to normalize ciliary function could ameliorate the symptoms of Huntington disease.

More information: Ciliogenesis is regulated by a huntingtin-HAP1-PCM1 pathway and is altered in Huntington disease:
[www.jci.org/articles/view/5755 ... b408131883a0d00ac557](http://www.jci.org/articles/view/5755...b408131883a0d00ac557)

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