

Yeast mimics severity of mutations leading to fatal childhood illness

December 22 2008

Scientists report that human gene mutations expressed in yeast cells can predict the severity of Batten Disease, a fatal nervous system disorder that begins during childhood. The new study published in *Disease Models* & *Mechanisms* (DMM), <u>dmm.biologists.org</u>, describes how the extent of changes in mutated cells paralleled the severity of symptoms seen in humans.

The initial, milder symptoms of Batten disease appear in children between ages 4 and 7. Children with this disorder (also known as juvenile neuronal ceroid lipfuscinosis, or JNCL) suffer vision loss and exhibit learning difficulties and behavioral changes. This is eventually followed by the appearance of seizures, and a devastating, progressive loss of mental and physical function, eventually leading to death before young adulthood.

Mutations in the gene CLN3 cause Batten Disease, but scientists do not fully understand the role of CLN3 in cell function. Thus, in order to learn more about this gene, researchers at the University College London created a variety of mutations based on CLN3 gene defects identified in Batten disease patients. They studied the effects of these mutations in a fission yeast protein highly similar to CLN3. The research team found that human mutations that caused a severe Batten disease progression likewise caused severe cell abnormalities in the yeast. Likewise, mutations found in mild cases of Batten disease resulted in less severe yeast cell changes.



Not only does this study help researchers understand the mechanism underlying Batten disease, but this yeast model can also be used to investigate therapeutic compounds to treat Batten disease and related illnesses.

Source: The Company of Biologists

Citation: Yeast mimics severity of mutations leading to fatal childhood illness (2008, December 22) retrieved 27 April 2024 from <u>https://phys.org/news/2008-12-yeast-mimics-severity-</u> mutations-fatal.html

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