CUL7 Gene

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Cullin 7: The CUL7 gene provides instructions for making a protein called cullin-7.

genes

1. Normal Function

This protein plays a role in the ubiquitin-proteasome system, which is the cell machinery that breaks down (degrades) unwanted proteins.

Cullin-7 helps assemble a complex known as an E3 ubiquitin ligase. This complex tags damaged and excess proteins with molecules called ubiquitin. Ubiquitin serves as a signal to specialized cell structures known as proteasomes, which attach (bind) to the tagged proteins and degrade them. The ubiquitin-proteasome system acts as the cell's quality control system by disposing of damaged, misshapen, and excess proteins. This system also regulates the level of proteins involved in several critical cell activities such as the timing of cell division and growth. In particular, cullin-7 is thought to help regulate proteins involved in the body's response to growth hormones, although its specific role in this process is unknown.

2. Health Conditions Related to Genetic Changes

2.1 3-M Syndrome

At least 73 mutations in the *CUL7* gene have been identified in people with 3-M syndrome, a disorder that causes skeletal anomalies including short stature (dwarfism) and unusual facial features. Most of these mutations substitute one protein building block (amino acid) for another amino acid in the cullin-7 protein or result in a cullin-7 protein that is abnormally short and nonfunctional.

Individuals in the Yakut population of the Russian province of Siberia with a variant of 3-M syndrome all have a particular mutation in both copies of the *CUL7* gene in each cell. This mutation replaces the amino acid glutamine with a premature stop signal in the instructions for making the cullin-7 protein (written as Gln1553Ter or Q1553X), leading to production of an abnormally short protein.

Mutations in the *CUL7* gene, including the Gln1553Ter mutation, prevent the cullin-7 protein from bringing together the components of the E3 ubiquitin ligase complex, interfering with the process of tagging unneeded proteins for

degradation. The body's response to growth hormones may be impaired as a result. However, the specific relationship between *CUL7* gene mutations and the signs and symptoms of 3-M syndrome are unknown.

3. Other Names for This Gene

- CUL7_HUMAN
- dJ20C7.5
- KIAA0076

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