CUL7 gene

cullin 7

Normal Function

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

Health Conditions Related to Genetic Changes

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.
Chromosomal Location
Cytogenetic Location: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1
Molecular Location: base pairs 43,037,617 to 43,053,945 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene
- CUL7_HUMAN
- dJ20C7.5
- KIAA0076

Additional Information & Resources
Educational Resources
  https://www.ncbi.nlm.nih.gov/books/NBK9957/#A1233

Clinical Information from GeneReviews
- 3-M Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1481

Scientific Articles on PubMed
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CUL7%5BTIAB%5D%29%29+OR+%28cullin+7%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Blanguage%5D+AND+human%5Bhuman+OR+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
- CULLIN 7
  http://omim.org/entry/609577
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CUL7.html
- ClinVar
- HGNC Gene Family: Cullins
  https://www.genenames.org/cgi-bin/genefamilies/set/1032
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21024
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9820
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q14999

Sources for This Summary

- OMIM: CULLIN 7
  http://omim.org/entry/609577
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22624670
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22156540
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23018678
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301654
Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16142236

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17675530
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2652813/


Reviewed: June 2018
Published: October 9, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services