ALMS1 gene

ALMS1 centrosome and basal body associated protein

Normal Function

The *ALMS1* gene provides instructions for making a protein whose function is unknown. Researchers believe that the protein may play a role in hearing, vision, regulation of body weight, and functions of the heart, kidney, lungs, and liver. It may also affect how the pancreas regulates insulin, a hormone that helps control levels of blood glucose, also called blood sugar.

The ALMS1 protein is present in most of the body's tissues, usually at low levels. Within cells, this protein is located in structures called centrosomes. Centrosomes play a role in cell division and the assembly of microtubules, which are proteins that transport materials in cells and help the cell maintain its shape. The ALMS1 protein is also found at the base of cilia, which are finger-like projections that stick out from the surface of cells. Almost all cells have cilia at some stage of their life cycle. Cilia are involved in cell movement and many different chemical signaling pathways. Based on its location within cells, researchers suggest that the ALMS1 protein might be involved in the organization of microtubules, the transport of various materials, and the normal function of cilia.

Health Conditions Related to Genetic Changes

Alström syndrome

More than 80 mutations in the *ALMS1* gene have been identified in people with Alström syndrome. Most of these mutations lead to the production of an abnormally small version of the ALMS1 protein that does not function properly. Researchers propose that a lack of normally functioning ALMS1 protein in the brain could lead to overeating. A loss of this protein in the pancreas may cause insulin resistance, a condition in which the body cannot use insulin properly. The combined effects of overeating and insulin resistance impair the body's ability to handle excess glucose, leading to diabetes and obesity (two common features of Alström syndrome). It is unclear how *ALMS1* mutations cause the other signs and symptoms of Alström syndrome. Researchers suspect that this condition is associated with malfunctioning cilia in many of the body's tissues and organs.

Other Names for This Gene

- ALMS1_HUMAN
- Alstrom syndrome 1
- Alstrom syndrome protein 1
- KIAA0328

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALMS1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7840[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALMS1%5BTIAB%5D%29+OR+%28KIAA0328%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ALMS1 CENTROSOME AND BASAL BODY ASSOCIATED PROTEIN; ALMS1 (https://omim.org/entry/606844)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7840)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALMS1[gene])

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Genomic Location

The *ALMS1* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

Last updated August 1, 2008