ARMC5 gene

armadillo repeat containing 5

Normal Function

The *ARMC5* gene provides instructions for making a protein about which little is known. It is found mainly in the fluid surrounding the cell nucleus (cytoplasm), and studies suggest that its function depends on interacting with other proteins. It is thought to act as a tumor suppressor, which means that it helps to prevent cells from growing and dividing too rapidly or in an uncontrolled way.

Health Conditions Related to Genetic Changes

Primary macronodular adrenal hyperplasia

At least 24 mutations in the *ARMC5* gene have been identified in people with primary macronodular adrenal hyperplasia (PMAH), a disorder that causes multiple lumps (nodules) to form in the adrenal glands, which are small hormone-producing glands located on top of each kidney. These nodules cause adrenal gland enlargement (hyperplasia) and result in production of higher-than-normal levels of the hormone cortisol. Cortisol normally helps maintain blood sugar (glucose) levels, protects the body from physical stress, and suppresses inflammation. Increased cortisol levels can lead to weight gain in the face and upper body, fragile skin, bone loss, fatigue, and other health problems, which often occur in people with PMAH.

People with PMAH caused by *ARMC5* gene mutations inherit one copy of the mutated gene in each cell. However, the condition develops only when affected individuals acquire a second mutation in the other copy of the *ARMC5* gene in certain cells of the adrenal glands. This second mutation is described as somatic. Instead of being passed from parent to child, somatic mutations are acquired during a person's lifetime and are present only in certain cells. Because somatic mutations are also required for PMAH to occur, some people who have inherited the altered *ARMC5* gene never develop the condition, a situation known as reduced penetrance.

The *ARMC5* gene mutations that cause PMAH are thought to impair the protein's tumor suppressor function, which allows the overgrowth of certain cells. It is unclear why this overgrowth is limited to the adrenal glands in people with PMAH.

Other Names for This Gene

- FLJ13063

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ARMC5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79798[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ARMC5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ARMADILLO REPEAT-CONTAINING PROTEIN 5; ARMC5 (https://omim.org/entry/615549)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/79798)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ARMC5[gene])

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Genomic Location

The *ARMC5* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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