ANKRD11 gene

ankyrin repeat domain containing 11

Normal Function

The *ANKRD11* gene provides instructions for making a protein called ankyrin repeat domain 11 (ANKRD11). As its name suggests, this protein contains multiple regions called ankyrin domains; proteins with these domains help other proteins interact with each other. The ANKRD11 protein interacts with certain proteins called histone deacetylases, which are important for controlling gene activity. Through these interactions, ANKRD11 affects when genes are turned on and off. For example, ANKRD11 brings together histone deacetylases and other proteins called p160 coactivators. This association regulates the ability of p160 coactivators to turn on gene activity. ANKRD11 may also enhance the activity of a protein called p53, which controls the growth and division (proliferation) and the self-destruction (apoptosis) of cells.

The ANKRD11 protein is found in nerve cells (neurons) in the brain. During embryonic development, ANKRD11 helps regulate the proliferation of these cells and development of the brain. Researchers speculate that the protein may also be involved in the ability of neurons to change and adapt over time (plasticity), which is important for learning and memory. ANKRD11 may function in other cells in the body and appears to be involved in normal bone development.

Health Conditions Related to Genetic Changes

KBG syndrome

Several *ANKRD11* gene mutations have been found to cause KBG syndrome, a condition characterized by large upper front teeth and other unusual facial features, skeletal abnormalities, and intellectual disability. Most of these mutations lead to an abnormally short ANKRD11 protein, which likely has little or no function. Reduction of this protein's function is thought to underlie the signs and symptoms of the condition. Because ANKRD11 is thought to play an important role in neurons and brain development, researchers speculate that a partial loss of its function may lead to developmental delay and intellectual disability in KBG syndrome. However, the mechanism is not fully known. It is also unclear how loss of ANKRD11 function leads to the skeletal features of the condition.

Another type of mutation that affects the *ANKRD11* gene, called 16q24.3 microdeletion,

deletes genetic material from chromosome 16 in a region designated q24.3. The deleted region typically removes the *ANKRD11* and *ZNF778* genes, although other nearby genes may also be affected. People with this type of mutation have similar signs and symptoms to those of KBG syndrome, including unusual facial features and intellectual disability. Many also have brain abnormalities and features of autism spectrum disorders, which are characterized by impaired communication and socialization skills. Some researchers think that these microdeletions are different enough from KBG syndrome to be considered a separate disorder, called 16q24.3 microdeletion syndrome.

Other Names for This Gene

- ANCO-1
- ANCO1
- ankyrin repeat domain-containing protein 11
- ankyrin repeat-containing cofactor 1
- LZ16
- nasopharyngeal carcinoma susceptibility protein
- T13

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ANKRD11 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=29123[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ANKRD11%5BTIAB%5D%29+OR+%28ankyrin+repeat+domain+11%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ANKYRIN REPEAT DOMAIN-CONTAINING PROTEIN 11; ANKRD11 (https://omim.org/entry/611192)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/29123)

- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ANKRD11[gene])

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Genomic Location

The *ANKRD11* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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