

NBEAL2 gene

neurobeachin like 2

Normal Function

The *NBEAL2* gene provides instructions for making a protein whose function is unclear. The protein appears to be critical for the normal development of platelets, which are small blood cells involved in blood clotting.

Platelets are produced in bone marrow, the spongy tissue in the center of long bones that produces most of the blood cells the body needs. Platelets are formed from large precursor cells known as megakaryocytes. Within these cells, the NBEAL2 protein is thought to play a role in the development of sacs called alpha-granules, which are the most abundant components of platelets. Alpha-granules contain growth factors and other proteins that are important for blood clotting and wound healing. In response to an injury that causes bleeding, the proteins stored in alpha-granules help platelets stick to one another to form a plug that seals off damaged blood vessels and prevents further blood loss.

Health Conditions Related to Genetic Changes

Gray platelet syndrome

At least 35 mutations in the *NBEAL2* gene have been found to cause gray platelet syndrome, a disorder associated with abnormal bleeding. Most people with gray platelet syndrome also develop a condition called myelofibrosis, which is characterized by the buildup of scar tissue (fibrosis) in the bone marrow that prevents it from making enough normal blood cells.

Mutations in the *NBEAL2* gene disrupt the normal production of alpha-granules in megakaryocytes. Without alpha-granules, platelets are abnormally large and fewer in number than usual (macrothrombocytopenia). The abnormal platelets also appear gray when viewed under a microscope, which gives this condition its name. A lack of alpha-granules impairs the clumping of platelets in response to injury, increasing the risk of abnormal bleeding. Myelofibrosis is thought to occur because the growth factors and other proteins that are normally packaged into alpha-granules leak out into the bone marrow. The proteins lead to fibrosis that affects the bone marrow's ability to make new blood cells.

Other Names for This Gene

- BDPLT4
- GPS
- KIAA0540
- neurobeachin-like 2
- neurobeachin-like protein 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NBEAL2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23218\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23218[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NBEAL2%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

- NEUROBEACHIN-LIKE 2; NBEAL2 (<https://omim.org/entry/614169>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23218>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NBEAL2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NBEAL2[gene]))

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Genomic Location

The *NBEAL2* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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