

## BDNF gene

brain derived neurotrophic factor

### Normal Function

The *BDNF* gene provides instructions for making a protein found in the brain and spinal cord called brain-derived neurotrophic factor. This protein promotes the survival of nerve cells (neurons) by playing a role in the growth, maturation (differentiation), and maintenance of these cells. In the brain, the BDNF protein is active at the connections between nerve cells (synapses), where cell-to-cell communication occurs. The synapses can change and adapt over time in response to experience, a characteristic called synaptic plasticity. The BDNF protein helps regulate synaptic plasticity, which is important for learning and memory.

The BDNF protein is found in regions of the brain that control eating, drinking, and body weight; the protein likely contributes to the management of these functions.

### Health Conditions Related to Genetic Changes

#### Opioid addiction

MedlinePlus Genetics provides information about Opioid addiction

#### WAGR syndrome

The *BDNF* gene is located in a region of chromosome 11 that is often deleted in a condition known as WAGRO syndrome. This condition is a variant of WAGR syndrome, which is a disorder that affects many body systems and is named for its main features: a childhood kidney cancer known as Wilms tumor, an eye problem called aniridia, genitourinary anomalies, and intellectual disability (formerly referred to as mental retardation). WAGRO syndrome also includes obesity. The deletions that cause WAGRO syndrome remove many genes from one copy of chromosome 11, including part or all of the *BDNF* gene. The loss of this gene is responsible for weight gain that begins in childhood in people with WAGRO syndrome.

People with WAGRO syndrome may be at greater risk of neurological problems such as intellectual disability and a developmental disorder called autism spectrum disorder that affects communication and social interaction than those with WAGR syndrome. It is unclear whether this increased risk is due to the loss of the *BDNF* gene or other nearby

genes.

### Other disorders

Certain common genetic variations (polymorphisms) in the *BDNF* gene have been associated with an increased risk of developing psychiatric disorders such as bipolar disorder, anxiety, and eating disorders.

Most studies have focused on the effects of a particular polymorphism in the *BDNF* gene. This variation alters a single protein building block (amino acid) in the protein, replacing the amino acid valine with the amino acid methionine at position 66 (written as Val66Met or V66M). This change impairs the protein's ability to function. Many studies report an association between the Val66Met polymorphism and psychiatric disorders; however, some studies have not supported these findings. It is unclear how changes in the *BDNF* gene are related to these disorders. A large number of genetic and environmental factors, most of which remain unknown, likely determine the risk of developing these complex conditions.

### **Other Names for This Gene**

- abrineurin
- ANON2
- BDNF\_HUMAN
- brain-derived neurotrophic factor
- BULN2
- neurotrophin

### **Additional Information & Resources**

#### Tests Listed in the Genetic Testing Registry

- Tests of BDNF ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=627\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=627[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28BDNF%5BTI%5D%29+OR+%28brain-derived+neurotrophic+factor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D%29%29%29>)

#### Catalog of Genes and Diseases from OMIM

- BRAIN-DERIVED NEUROTROPHIC FACTOR; BDNF (<https://omim.org/entry/1135>)

05)

- BULIMIA NERVOSA, SUSCEPTIBILITY TO; BULN (<https://omim.org/entry/607499>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/627>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=BDNF\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=BDNF[gene]))

### **References**

- Han JC, Liu QR, Jones M, Levinn RL, Menzie CM, Jefferson-George KS, Adler-Wailes DC, Sanford EL, Lacbawan FL, Uhl GR, Rennert OM, Yanovski JA. Brain-derived neurotrophic factor and obesity in the WAGR syndrome. *N Engl J Med*. 2008 Aug 28;359(9):918-27. doi: 10.1056/NEJMoa0801119. Erratum In: *N Engl J Med*. 2008 Sep 25;359(13):1414. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18753648>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2553704/>)
- Rodriguez-Lopez R, Perez JM, Balsera AM, Rodriguez GG, Moreno TH, Garcia deCaceres M, Serrano MG, Freijo FC, Ruiz JR, Angueira FB, Perez PM, Estevez MN, Gomez EG. The modifier effect of the BDNF gene in the phenotype of the WAGRO syndrome. *Gene*. 2013 Mar 10;516(2):285-90. doi: 10.1016/j.gene.2012.11.073. Epub 2012 Dec 21. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23266638>)
- Sears C, Markie D, Olds R, Fitches A. Evidence of associations between bipolar disorder and the brain-derived neurotrophic factor (BDNF) gene. *Bipolar Disord*. 2011 Nov-Dec;13(7-8):630-7. doi: 10.1111/j.1399-5618.2011.00955.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22085476>)
- Shinawi M, Sahoo T, Maranda B, Skinner SA, Skinner C, Chinault C, Zascavage R, Peters SU, Patel A, Stevenson RE, Beaudet AL. 11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. *Am J Med Genet A*. 2011 Jun;155A(6):1272-80. doi: 10.1002/ajmg.a.33878. Epub 2011 May 12. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21567907>)

### **Genomic Location**

The *BDNF* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

**Last updated March 1, 2013**