

# PHF21A Gene

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PHD finger protein 21A

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## 1. Introduction

The *PHF21A* gene (also known as *BHC80*) provides instructions for making a protein involved in a process called histone demethylation, which helps control (regulate) gene activity. Histones are structural proteins that attach (bind) to DNA and give chromosomes their shape. The removal of a molecule called a methyl group from histones (histone demethylation), helps turn off (repress) certain genes. The PHF21A protein binds to histones that have already been demethylated, which researchers speculate helps keep the histone demethylated and the genes turned off. The PHF21A protein appears to be particularly important in regulating genes involved in development of nerve cells in the brain and structures of the face.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Potocki-Shaffer syndrome

A genetic change resulting in the deletion of the *PHF21A* gene causes a condition called Potocki-Shaffer syndrome. People with this condition have enlarged openings in two bones that make up much of the top and sides of the skull (enlarged parietal foramina) and multiple noncancerous bone tumors (osteochondromas). Other signs and symptoms seen in some people with Potocki-Shaffer syndrome include intellectual disability, developmental delay, distinctive facial features, vision problems, and defects in the heart, kidneys, and urinary tract.

Potocki-Shaffer syndrome (also called proximal 11p deletion syndrome) is caused by a deletion of genetic material from the short (p) arm of chromosome 11. In people with this condition, a loss of the *PHF21A* gene within this region is responsible for intellectual disability and distinctive facial features. The deletion likely leads to a reduction in the amount of PHF21A protein. It is thought that the resulting disruption of histone demethylation alters the activity of genes involved in neuronal and facial development, leading to intellectual disability and distinctive facial features. The loss of other genes in the same region of chromosome 11, *ALX4* and *EXT2*, underlie the enlarged parietal foramina and multiple osteochondromas, respectively. The loss of additional genes in the deleted region likely contributes to the other features of Potocki-Shaffer syndrome.

## 3. Other Names for This Gene

- BHC80
- BHC80a
- BM-006
- BRAF35-HDAC complex protein BHC80
- BRAF35/HDAC2 complex (80 kDa)
- KIAA1696

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