# CHD7 Gene

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chromodomain helicase DNA binding protein 7

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## 1. Normal Function

The *CHD7* gene provides instructions for making a protein called chromodomain helicase DNA binding protein 7. This protein is found in many parts of the body before birth, including the eye, the inner ear, and the brain. In the brain, the CHD7 protein is active in several areas, including a bundle of nerve cells (neurons) called the olfactory bulb that is critical for the perception of odors.

The CHD7 protein belongs to a family of proteins that are thought to play a role in the organization of chromatin. Chromatin is the complex of DNA and protein that packages DNA into chromosomes. The CHD7 protein regulates the activity (expression) of several other genes through a process known as chromatin remodeling. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. When DNA is tightly packed, gene expression is lower than when DNA is loosely packed. Researchers are working to determine which genes the CHD7 protein regulates.

## 2. Health Conditions Related to Genetic Changes

### 2.1. CHARGE syndrome

Mutations in the *CHD7* gene cause CHARGE syndrome, a disorder that affects many areas of the body. CHARGE is an abbreviation for several of the features common in the disorder: coloboma, heart defect, atresia choanae (also known as choanal atresia), growth retardation, genital abnormality, and ear abnormality. More than 600 mutations that can cause CHARGE syndrome have been identified, and they occur throughout the *CHD7* gene. Most of these mutations lead to the production of an abnormal CHD7 protein that is broken down prematurely. Shortage of this protein is thought to disrupt chromatin remodeling and the regulation of gene expression. Changes in gene expression during embryonic development likely cause the signs and symptoms of CHARGE syndrome.

### 2.2. Kallmann Syndrome

More than 50mutations in the *CHD7* gene have been identified in people with Kallmann syndrome, a disorder characterized by the combination of hypogonadotropic hypogonadism (a condition affecting the production of hormones that direct sexual development) and an impaired sense of smell. Mutations in this gene account for 5 to 10 percent of all cases of Kallmann syndrome.

Many people with Kallmann syndrome caused by a *CHD7* gene mutation have some of the features of CHARGE syndrome (described above), such as abnormally shaped ears and hearing loss. However, the signs and symptoms tend to be much less severe. Researchers suspect that Kallmann syndrome resulting from a *CHD7* gene mutation may actually represent a mild form of CHARGE syndrome.

Most of the *CHD7* gene mutations that cause Kallmann syndrome alter single protein building blocks (amino acids) in the CHD7 protein. Studies suggest that these mutations have a less severe effect on protein function than those that cause CHARGE syndrome. The altered protein affects the development of the olfactory bulb, which impairs the sense of smell. It also disrupts the development of certain neurons needed for the production of sex hormones, which interferes with normal sexual development.

### 2.3. Coloboma

Coloboma

## 3. Other Names for This Gene

- CHD7_HUMAN
- FLJ20357
- FLJ20361
- IS3
- KIAA1416

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