

# 7q11.23 Duplication Syndrome

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7q11.23 duplication syndrome is a condition that can cause a variety of neurological and behavioral problems as well as other abnormalities.

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## 1. Introduction

People with 7q11.23 duplication syndrome typically have delayed development of speech and motor skills such as crawling and walking. Speech problems and abnormalities in the way affected individuals walk and stand may persist throughout life. People with this condition may also have weak muscle tone (hypotonia) and abnormal movements, such as involuntary movements of one side of the body that mirror intentional movements of the other side. About one-fifth of people with 7q11.23 duplication syndrome experience seizures.

Intellectual development varies widely in 7q11.23 duplication syndrome. The majority of people with this condition have low-average to average intelligence. Intellectual disability or borderline intellectual ability occur in about one-third of affected individuals. Rarely, people with this disorder have above-average intelligence.

Behavioral problems associated with this condition include anxiety disorders (such as social phobias and selective mutism, which is an inability to speak in certain circumstances), attention-deficit/hyperactivity disorder (ADHD), physical aggression, excessively defiant behavior (oppositional disorder), and autistic behaviors that affect communication and social interaction.

Approximately half of individuals with 7q11.23 duplication syndrome have enlargement (dilatation) of the blood vessel that carries blood from the heart to the rest of the body (the aorta); this enlargement can get worse over time. Aortic dilatation can lead to life-threatening complications if the wall of the aorta separates into layers (aortic dissection) or breaks open (ruptures).

People with 7q11.23 duplication syndrome can have characteristic features of the head and face, including a large head (macrocephaly) that is flattened in the back (brachycephaly), a broad forehead, straight eyebrows, and deep-set eyes with long eyelashes. The nose may be broad at the tip with the area separating the nostrils attaching lower than usual on the face (low insertion of the columella), resulting in a shortened area between the nose and the upper lip (philtrum). A high arch in the roof of the mouth (high-arched palate) and ear abnormalities may also occur. These features may be mild and not recognized in some affected individuals.

## 2. Frequency

The prevalence of this disorder is estimated to be 1 in 7,500 to 20,000 people.

## 3. Causes

7q11.23 duplication syndrome results from an extra copy of a region on the long (q) arm of chromosome 7 in each cell. This region is called the Williams-Beuren syndrome critical region (WBSCR) because its deletion causes a different disorder called Williams syndrome, also known as Williams-Beuren syndrome. The region, which is 1.5 to 1.8 million DNA base pairs (Mb) in length, includes 26 to 28 genes.

Extra copies of several of the genes in the duplicated region, including the *ELN* and *GTF2I* genes, likely contribute to the characteristic features of 7q11.23 duplication syndrome. Researchers suggest that an extra copy of the *ELN* gene in each cell may be related to the increased risk for aortic dilatation in 7q11.23 duplication syndrome. Studies suggest that an extra copy of the *GTF2I* gene may be associated with some of the behavioral features of the disorder. However, the specific causes of these features are unclear.

Researchers are studying additional genes in the duplicated region, but none have been definitely linked to any of the specific signs or symptoms of 7q11.23 duplication syndrome.

### 3.1. The genes and chromosome associated with 7q11.23 duplication syndrome

- *ELN*
- *GTF2I*
- chromosome 7

## 4. Inheritance

7q11.23 duplication syndrome is considered to be an autosomal dominant condition, which means one copy of chromosome 7 with the duplication in each cell is sufficient to cause the disorder. Most cases result from a duplication that occurs during the formation of reproductive cells (eggs and sperm). These cases occur in people with no history of the disorder in their family. In about a quarter of cases, an affected person inherits the chromosome with a duplicated segment from a parent. Typically, the parent has subtle signs and symptoms related to the duplication and is not diagnosed until after the disorder is recognized in the child.

## 5. Other Names for This Condition

- 7q11.23 microduplication syndrome
- chromosome 7q11.23 duplication
- chromosome 7q11.23 duplication syndrome
- dup(7)(q11.23)
- Somerville-Van der Aa syndrome
- trisomy 7q11.23
- WBS duplication syndrome
- Williams-Beuren region duplication syndrome

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