Ring Chromosome 14 Syndrome

Subjects: Genetics
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Definition

Ring chromosome 14 syndrome is a condition characterized by seizures and intellectual disability.

1. Introduction

Recurrent seizures (epilepsy) develop in infancy or early childhood. In many cases, the seizures are resistant to treatment with anti-epileptic drugs. Most people with ring chromosome 14 syndrome also have some degree of intellectual disability or learning problems. Development may be delayed, particularly the development of speech and of motor skills such as sitting, standing, and walking.

Additional features of ring chromosome 14 syndrome can include slow growth and short stature, a small head (microcephaly), puffy hands and/or feet caused by a buildup of fluid (lymphedema), and subtle differences in facial features. Some affected individuals have problems with their immune system that lead to recurrent infections, especially involving the respiratory system. Abnormalities of the retina, the specialized tissue at the back of the eye that detects light and color, have also been reported in some people with this condition. These changes typically do not affect vision. Major birth defects are rarely seen with ring chromosome 14 syndrome.

2. Frequency

Ring chromosome 14 syndrome appears to be a rare condition, although its prevalence is unknown. More than 50 affected individuals have been reported in the medical literature.

3. Causes

Ring chromosome 14 syndrome is caused by a chromosomal abnormality known as a ring chromosome 14, sometimes written as r(14). A ring chromosome is a circular structure that occurs when a chromosome breaks in two places and its broken ends fuse together. People with ring chromosome 14 syndrome have one copy of this abnormal chromosome in some or all of their cells.

Researchers believe that several critical genes near the end of the long (q) arm of chromosome 14 are lost when the ring chromosome forms. The loss of these genes is likely responsible for several of the major features of ring chromosome 14 syndrome, including intellectual disability and delayed development. Researchers are still working to determine which missing genes contribute to the signs and symptoms of this disorder.

Epilepsy is a common feature of ring chromosome syndromes, including ring chromosome 14. There may be something about the ring structure itself that causes epilepsy. Seizures may occur because certain genes on the ring chromosome 14 are less active than those on the normal chromosome 14. Alternately, seizures might result from instability of the ring chromosome in some cells.

3.1. The Chromosome Associated with Ring Chromosome 14 Syndrome

- chromosome 14

4. Inheritance

Ring chromosome 14 syndrome is almost never inherited. A ring chromosome typically occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early embryonic development. In some cases, the ring chromosome is present in only some of a person's cells. This situation is known as mosaicism.

Most affected individuals have no history of the disorder in their families. However, at least two families have been reported in which a ring chromosome 14 was passed from a mother to her children.

5. Other Names for This Condition

- ring 14
- ring 14 syndrome
- ring chromosome 14

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Keywords

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