

Ring chromosome 20 syndrome

Description

Ring chromosome 20 syndrome is a condition that affects the normal development and function of the brain. The most common feature of this condition is recurrent seizures (epilepsy) in childhood. The seizures may occur during the day or at night during sleep. They are described as partial seizures because they affect only one area of the brain, a region called the frontal lobe. In many cases, the seizures are complex and resistant to treatment with anti-epileptic drugs. Prolonged seizure episodes known as non-convulsive status epilepticus also appear to be characteristic of ring chromosome 20 syndrome. These episodes involve confusion and behavioral changes.

Most people with ring chromosome 20 syndrome also have intellectual disabilities and behavioral difficulties. Although these problems can appear either before or after the onset of epilepsy, they tend to worsen after seizures develop. Major birth defects and differences in facial features can occur in people with ring chromosome 20 syndrome, though these are rare.

Frequency

Ring chromosome 20 syndrome appears to be a rare condition, although its prevalence is unknown. More than 200 affected individuals have been reported in the medical literature.

Causes

Ring chromosome 20 syndrome is caused by a chromosomal abnormality known as a ring chromosome 20 or r(20). 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 20 syndrome have one copy of this abnormal chromosome in some or all of their cells.

It is not well understood how the ring chromosome causes the signs and symptoms of this syndrome. In some affected individuals, genes near the ends of chromosome 20 are deleted when the ring chromosome forms. Researchers suspect that the loss of these genes may be responsible for epilepsy and other health problems. However, most affected individuals do not have these gene deletions. In these people, the ring chromosome may change the activity of certain genes on chromosome 20, or the

chromosome may be unable to copy (replicate) itself normally during cell division. Researchers are still working to determine the precise relationship between the ring chromosome 20 and the characteristic features of the syndrome.

[Learn more about the chromosome associated with Ring chromosome 20 syndrome](#)

- chromosome 20

Inheritance

Ring chromosome 20 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. Often, 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, there have been a few reports of a ring chromosome 20 being passed from a parent to a child.

Other Names for This Condition

- R(20) syndrome
- Ring 20 syndrome
- Ring chromosome 20
- Ring chromosome 20 epilepsy syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Ring chromosome 20 syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265482/>)

Genetic and Rare Diseases Information Center

- Ring chromosome 20 syndrome (<https://rarediseases.info.nih.gov/diseases/1334/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Ring+Chromosomes%5BMAJR%5D%29+AND+%28%28ring+chromosome+20+syndrome%5BTIAB%5D%29+OR+%28ring+chromosome+20%5BTIAB%5D%29+OR+%28ring+20+syndrome%5BTIAB%5D%29+OR+%28ring+chromosome+20+epilepsy+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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