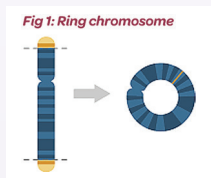




Ring Chromosome 20 Syndrome

Also known as: r(20) syndrome, r(20), Ring 20, RC20

Overview



In Ring Chromosome 20 Syndrome one of the two copies of chromosome 20 has formed a ring rather than the typical linear chromosome structure (figure 1). Why the tips of the chromosome join together to form the ring is not understood and how

the formation of the ring affects the function of genes packaged on chromosome 20 is also not understood. The consequence of the ring formation is a difficult to control epilepsy, typically with onset in early childhood and associated with intellectual disability and behaviour problems which may be progressive. Sometimes the behavioural problems may present before the epilepsy. The diagnosis of r(20) syndrome is often delayed because physicians may not consider checking chromosomes in people who have no intellectual disability before the epilepsy begins.

Another reason for diagnostic delay is that modern methods of testing for chromosome abnormalities, such as chromosome microarray, are better at picking up small changes in chromosomes but can miss the formation of rings. An older test (a karyotype test) where the chromosomes are looked at under a microscope must be requested to diagnose the condition. Not all the cells in the body may have a ring chromosome. When only a percentage of cells have a ring chromosome 20 this is called mosaicism. In general, a higher proportion of abnormal cells is associated with earlier age of onset of seizures, but this not true for everyone. If all the cells in the body have a ring chromosome 20 the individual tends to be more severely affected. r(20) syndrome is not usually inherited, but affected individuals should seek genetic counselling if wishing to start a family of their own.

1. How common is r(20) syndrome?

This is a rare disorder and we do not know how common it is. There are likely to be people with epilepsy in which the syndrome remains undiagnosed or misdiagnosed.

2. When do symptoms first appear?

Usually seizures begin in early childhood, typically around the time children start school however symptoms may also present

earlier in childhood, in adolescence or early adult life. Unlike many chromosomal disorders people with r(20) syndrome have a normal appearance and do not have intellectual disability before the epilepsy onset. There is usually a sudden onset of seizures without any clear trigger. Many people experience a change in behaviour before or around the seizure onset, manifested by poor attention and concentration, impulsivity and other behavioural problems

3. What are the types of seizure(s) seen in r(20) syndrome?

Several different seizure types may be seen. For accepted definitions of seizure types see EpiCARE seizure types leaflet or www.epilepsydiagnosis.org. Features characteristic of r(20) syndrome are:

Focal seizures with impaired awareness

- At onset these are often from sleep. The person may rouse suddenly, sit up and have brief abnormal movements (stiffening or abnormal postures) of their arms and legs and may shout out before settling to sleep. These may occur several times a night and can be mistaken for night terrors.
- Events in the daytime where the individual appears frightened, confused and does not respond normally.
- A feature characteristic of focal seizures in r(20) syndrome is frightening hallucinations. Examples include seeing sharks swimming above them, fire, spiders or large black holes. These may be mistaken for non-epileptic events.
- Sometimes people may have clusters of seizures with brief periods of apparently normal awareness between several events.

Tonic clonic seizures – These can occur in isolation or evolve from focal seizures

Non-convulsive status epilepticus – These are prolonged focal seizures with impairment of awareness which may last many minutes to hours (rarely days). They often occur in the late afternoon and early evening.

Other seizure types include **myoclonic seizures** (rapid jerks of muscles) or **atonic seizures** (loss of muscle tone) – both seizure types may result in sudden drops or falls.

4. Is r(20) syndrome linked to any other epilepsy syndromes?

Epilepsies can be defined as syndromes based on the different seizure types, EEG patterns, age of onset or based on the cause, if known, as well as associated co-morbidities (see other problems below). r(20) syndrome is an epilepsy syndrome in its own right as it has characteristic features with a specific genetic cause.

5. How frequent are seizures typically in r(20) syndrome?

Seizures may become very frequent with multiple events per day.

6. How may seizures change over time?

Nocturnal focal seizures are often the first seizure type. People with r(20) syndrome can go through periods where they have multiple very difficult to control seizures on a daily basis and this may be associated with significant intellectual and behavioural decline. These periods tend to be more of a problem in the first few years after presentation. Later in the course of epilepsy the seizures may become less frequent and may change over time with a more predictable pattern specific for that individual. Complete seizure freedom for prolonged periods (years) appears to be exceptional with most people having seizures in adult life.

7. What other problems apart from epilepsy, affect people with r(20) syndrome?

Until the onset of seizures childhood development appears to be normal, however there often follows a rapid decline in intellectual function. Skills previously attained may be lost. This decline in ability varies from losing some skills but remaining in the normal range for intellectual function, to a significant loss in skills including difficulties with mobility and loss of speech and language functions. When this decline is associated with epilepsy it is called an epileptic encephalopathy. When very severe, r(20) syndrome may manifest in some people as a form of childhood dementia.

Support is likely to be required at school/college or even in the workplace. Behavioural issues such as bouts of aggression before and/or after a seizure are commonly reported. Individuals may also receive a diagnosis of autism. Predicting how severe these learning and behaviour problems will be in the future is not possible at the onset of the epilepsy.

8. What are the treatment options for r(20) syndrome?

Seizures do not typically respond to treatment and there is currently no recommended treatment for r(20) syndrome. Treatment usually comprises use of anti-epileptic drugs (AED's), although Vagus Nerve Stimulation (VNS) and/or Ketogenic Dietary Therapy (KDT) have proven useful in some cases. r(20) syndrome is not suitable for brain surgery.

9. What is the emergency protocol for seizures?

Your doctor may advise special treatment for emergency situations as prolonged seizures may be dangerous to health and must be treated immediately. It is important that every person has an individualised treatment plan for emergencies.

10. What could I ask my doctor or specialist epilepsy nurse about?

- Safety advice especially re the 'confusional state' associated with episodes of NCSE
- A personalised rescue medication plan for prolonged or cluster seizures
- The side effects of medication particularly when changing treatment
- Genetic counselling
- Liaison with school or college for support during education
- Patient, carer & employer support requirements including neuropsychological evaluation, guidance, potential psychiatric support
- Sudden Unexpected Death in Epilepsy (SUDEP) risk management



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