Ring Chromosome 20 (R20)

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For further information about epilepsy or anything mentioned in this factsheet, please contact the Epilepsy Helpline freephone 0808 800 5050 or helpline@epilepsy.org.uk.

What is a syndrome?
A syndrome is a group of signs and symptoms that, added together, suggest a particular medical condition. In epilepsy, examples of these signs and symptoms would be things like the age at which seizures begin, the type of seizures, whether the child is male or female and whether they experience difficulties with learning.

Ring Chromosome 20 (R20)
This is a rare condition which causes epilepsy in children. Since it was first reported in 1972 there have been over 50 cases reported worldwide. It is caused by an abnormality in chromosome number 20. Chromosomes are structures within each cell in the body which hold our genetic material. Each human being has 23 pairs of chromosomes. That is 46 in total. When viewed down a microscope, chromosomes usually look fairly straight. In R20 a little piece of genetic material is missing from each end of one of the number 20 chromosomes and the ends fuse together to form a ring. This happens to the developing baby very early in pregnancy. It happens sporadically (by chance) and therefore does not affect more than one child in each family.

Symptoms
The first symptom of R20 is usually epilepsy and can start at any time from day one of life until 17 years. Patients may have mild learning difficulties before the start of epilepsy. There are a variety of seizure types. *Nocturnal (night time) seizures are frequent and may not be noticed. *Complex partial seizures can take on many forms including head turning, jerking of one or more limb and episodes of altered consciousness with repetitive mouth movements. *Tonic-clonic and absence seizures can also happen. People with R20 seem particularly prone to episodes of *non-convulsive status epilepticus (NCSE). This is happens when there is continuous abnormal electrical activity from the brain and is associated with a change of behaviour, ranging from mild confusion to a severe altered state of consciousness. These can last from 30 minutes to days or even weeks in some cases. The start of epilepsy in R20 often comes before a decline in the person’s learning abilities and behaviour. Behaviour problems may be severe and can deteriorate to the extent that many children are referred to a child psychiatric service before the diagnosis is made.

Diagnosis
Examining the child’s chromosomes under a microscope makes the diagnosis. This involves a blood test.
Severe epilepsy with periods of non-convulsive status epilepticus (NCSE) associated with the start of learning and behaviour difficulties may suggest a diagnosis of Ring Chromosome 20. Some electroencephalogram (EEG) features seem to be fairly common in R20 and may again suggest the diagnosis. Unfortunately, because the condition is so rare, the diagnosis is often delayed. Brain scans are normal in children with R20.

Treatment
The epilepsy associated with R20 syndrome is difficult to control. No single anti-epileptic drug has been shown to be particularly effective in this condition and multiple drugs either in isolation or in combination may be tried. There are a small number of single case reports suggesting that vagal nerve stimulation may be effective in R20. Periods of NCSE are also difficult to treat. Short-term treatment with benzodiazepines (clobazam, midazolam, diazepam) or steroid medications (prednisolone) may be used to treat these episodes but the person often has further episodes of NCSE when these medications have been withdrawn.

Prognosis (outlook)
The long-term prognosis of R20 is variable and depends on the person, the severity of the epilepsy and the associated learning and behavioural problems. More studies are needed to clarify the longer-term outlook for people with R20.

For information on seizure types, anti-epileptic medication, vagal nerve stimulation and general aspects of epilepsy please contact the Epilepsy Helpline freephone 0808 800 5050.

The Ring Chromosome 20 Foundation, One Angel Court, London EC2R 7HJ, www.ring-chromosome-20.org, email info@ring-chromosome-20.org

Contact a Family, 209-211 City Road, London, EC1V 1JN, telephone 0808 808 3555, www.cafamily.org.uk

Epilepsy Helpline
freephone 0808 800 5050
text 07797 805 390
helpline@epilepsy.org.uk
www.epilepsy.org.uk

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If you have any comments you would like to make about this fact sheet, please contact us.

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Contact details
Epilepsy Action, New Anstey House, Gate Way Drive, Yeadon, Leeds LS19 7XY.
Tel: 0113 210 8800. Fax: 0113 391 0300.
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