WHO ARE RING 20 RESEARCH SUPPORT UK?

Ring20 Research and Support UK has been set up to support families, individuals and professionals who are affected by, or who come into contact with r(20).

We operate from the UK (England) but are happy to extend our support to the many families affected by r(20) around the world and the health professionals that look after them.

Our mission is to raise funds for future research studies by way of member fundraising and seeking out opportunities for grants and/or corporate sponsorship.

We aim to represent r(20) patients and their families as a patient cohort to aid vital research, which may lead to improved quality of life and improved outcomes for those living with, or affected by r(20).

Membership provides a support network for our families to connect with each other in an otherwise isolated environment. We hope that by sharing experiences and knowledge we can help each other.

HOW CAN YOU HELP?

To achieve our mission we need to identify as many r(20) families as possible, to offer them our support in response for their input; information sharing, experience of living with r(20), treatment options, fundraising and more.

Membership is free and provides you with access to regular updates on our progress and activities and our patient support group forums (one for families and one exclusively for individuals with the condition) both on Facebook:

Family Support Group Forum:
www.facebook.com/groups/798475916833994/

Patient Support Group Forum:
www.facebook.com/groups/229551470569368/

We ask that all families, individuals and professionals that wish to engage with us become members, in order that they may jointly contribute to our cause, by virtue of time, money or information (no matter how large or small).

☐ Yes I would like to join Ring 20 Research Support UK

☐ Yes, I would like to make a donation to Ring 20 Research Support UK (Gift Aid)

Enclosed is my Gift of: £5 ☐ £10 ☐ £25 ☐

£50 ☐ £100 ☐

Other _____

Please make my donation in honour of

_____ (optional)

Name: _____

Address: _____

City: _____ County: _____

Postcode: _____ Tel: _____

e-mail: _____

Make cheques payable to Ring20 Research Support UK
Enclose form and cheque and mail to:
The Secretary, 26 Headley Chase, Warley, Brentwood, Essex CM14 5BN
Thank you

www.ring20researchsupport.co.uk

26 Headley Chase, Warley, Brentwood,
Essex CM14 5BN England
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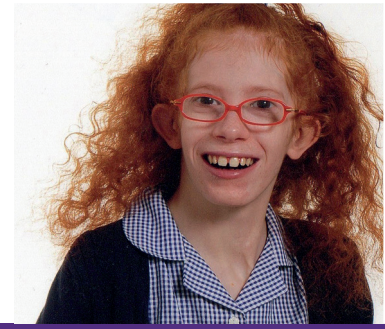
Do You Know About Ring Chromosome 20 Syndrome?








WHAT IS R(20) SYNDROME?

Ring chromosome 20 r(20) epilepsy syndrome was first categorised in literature as a genetic syndrome in 1976 by Borgaonkar and colleagues at John Hopkins University in the United States. Since then many cases have been reported in the literature worldwide.

Estimates of the prevalence of this rare disorder are as yet unknown. However with more awareness and increased clinical recognition more cases will undoubtedly be discovered.

This condition is typically characterised by drug-resistant focal epilepsy (previously called complex partial), behavioural issues and varying degree of intellectual impairment. Two distinct clinical subtypes (mosaic and non-mosaic) can be recognised clinically and confirmed by chromosomal studies. Chromosomal karyotype with screening for mosaicism (to identify cases where the ring is not in every cell) is considered to be the best diagnostic test for identification of this disorder.

R(20) Syndrome appears to be a pan-ethnic and nongender specific disorder with the recurrent risk being very low as the chromosomal ring formation is not usually inherited but rather occurs from a spontaneous, or 'de novo' event. However a few familial cases have been reported in literature.

Ring formation can occur at different stages of development, and this dictates what percentage of cells contain the ring chromosome. In cases where the ring forms early, in germ cells (egg or sperm), all cells will contain a ring chromosome and a person will be non-mosaic. In cases where a ring chromosome occurs postzygotically (union of the egg and sperm to create the early embryo), only a percentage of cells will contain the ring chromosome and a person will be mosaic.

Research has shown that there is a correlation between mosaicism and r(20) clinical features, where mosaic patients tend to have later seizure onset and a lower incidence of additional symptoms when compared to patients who are non-mosaic for r(20).

WHAT IS THE COURSE OF R(20)?

The course of r(20) largely depends on the degree of mosaism and is variable from child to child. Seizures may not present until up to four years after birth. Up until that point development appears to be normal. Epilepsy is a constant feature of this syndrome and is seen in almost 100% of the cases and usually starts in early childhood with many cases being intractable and drug resistant (difficult to treat).

Seizures are primarily focal (complex partial) in type and can be accompanied by episodes of altered consciousness with staring. Periods of intense fear and prolonged confused state lasting several minutes are also noted (non-convulsive status epilepticus). Features of frontal lobe epilepsy are often reported and subtle nocturnal seizures (SNS) and nocturnal frontal lobe seizures (SNFLS) are also recognised.

Behavioural problems in children have often been reported in the literature and can vary from minor concentration and attention issues to high levels of activity often associated with poor seizure control. Some individuals may have profound learning difficulties while others perform well with well controlled seizures.

Abnormal physical / dysmorphic features are often lacking which can lead to misdiagnosis or delayed diagnosis.

WHY DIAGNOSING R(20) MATTERS?

There are many reasons why patients have seizures, both genetic and non-genetic. Finding the cause is enormously helpful to physicians to guide treatment, identify other clinical problems that patients may be at risk for, and provide prognostic information. It is also very emotionally beneficial for a family to be able to have a diagnosis for their child. Since r(20) is diagnosed by chromosome analysis, which is not a routine procedure for most patients with seizures, this disorder is likely under-diagnosed. This syndrome can be confused with Lennox Gastaut Syndrome (LGS) and other epilepsy syndromes with frontal lobe features. The electroencephalogram (EEG) may have distinguishing features in sleep but these are not always recognized.

DIAGNOSING R(20):

Appropriate Diagnostic Testing Procedure

R(20) is usually diagnosed by cytogenetic (chromosome) analysis from a small blood sample. The chromosomes from the cells in the blood are examined and the ring is usually obvious under a microscope. The diagnosis can also be made from skin cells using the same cytogenetic technique.

Chromosomal microarray testing will identify deletions that may occur when the ring is formed, but this is only true for non-mosaic r(20) patients. For a diagnosis, it remains necessary to look at chromosomes through a microscope to see their shape. However, additional molecular genetic tests such as FISH and microarrays from the same blood sample can show more precisely how the ends of the chromosome have joined and whether any genetic material was deleted or duplicated in the process. The r(20) may also be analysed from skin cells, but this is rare because obtaining a skin biopsy for the test is more invasive and most physicians and patients prefer to obtain a blood sample in most cases.

What to do if you suspect r(20)

If you suspect r(20), cytogenetic (chromosome) confirmation of the diagnosis is essential. Discuss chromosome analysis with mosaic screening with your neurologist. Newer genetic testing such as chromosomal microarray will not diagnose r(20) in most cases.

HOW IS R(20) SYNDROME TREATED?

The main-stay of treatment is antiepileptic drug (AED) therapy. There are no comparative studies that have been done and in recent published literature no one drug seems to be better than another. Patients are frequently subjected to multiple AEDs (polytherapy), with limited seizure improvement. It is well known that poly-therapy increases the risk of drug side effects.

Both Vagus Nerve Stimulation (VNS) therapy and the ketogenic diet have been reported to be beneficial. Both treatments are now well established in terms of safety and efficacy in the treatment of drug-resistant epilepsies. More research and reported case series are required in order to better evaluate alternative treatment therapies for r(20).