CAN YOU HELP US?

If you have a research idea, or are interested in undertaking research into r(20) syndrome then we would love to hear from you. We welcome expressions of interest from all stakeholders including: researchers, academics, clinicians, geneticists, pharma, industry and biotech and more,

If you are a funder who can support our work in illuminating the rare reality of living with r(20) syndrome through awareness campaigns, improving understanding of the condition, or by helping us to work with partners to find more effective treatments, then please do get in touch.

ring 20
research & support

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Let's Connect

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Why research r(20)?

ring 20
research & support

Information and Support services
Making connections, Supporting families

The facts

What is r(20) syndrome?

Ring Chromsome 20 Syndrome [r(20) syndrome] is an ultra-rare epilepsy. There are only 200 cases reported in medical literature worldwide.
Individuals with r(20) syndrome experience debilitating regular seizures, often multiple seizures every day of a variety of seizure types, accompanied by cognitive decline and challenging behaviours following seizure onset. Unlike many chromosomal disorders, symptoms do not typically present from birth.

Who are Ring20?

...supporting 60% of patient families globally

At Ring20 Research and Support (Ring20) our mission is to provide support for individuals, families and healthcare professionals who are affected by, or who come into contact with r(20) syndrome and to continuously foster interest and promote opportunities for research to increase understanding and improve outcomes for all.

The unmet needs

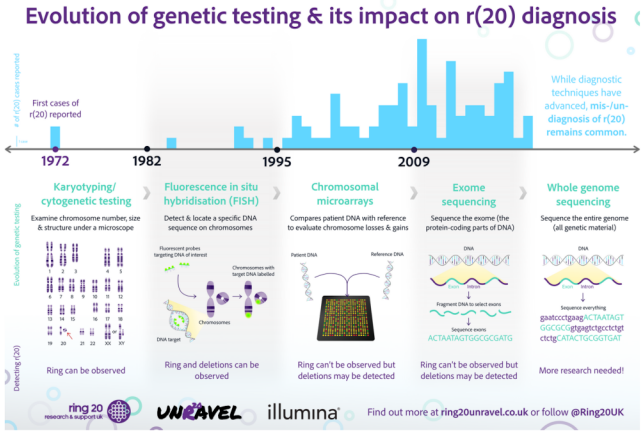
- No patient registries
- No natural history studies
- No cell/animal models
- No clinical trials (ever)
- No clinical guidelines. or consensus statements
- No recommended treatment

The time is ripe for investment.

Research challenges

• Diagnosis

r(20) syndrome is likely under-reported and under diagnosed, and through our membership we have witnessed a global decline in diagnoses, but WHY? Next Generation Sequencing cannot (yet) identify the structural change in the chromosome.



• Treatment

Individuals are treated on a 'trial and error' basis, often subjected to a cocktail of anti-seizure medications /treatments with limited efficacy on seizure control, whilst the side effects further impair Quality of Life.

• Care

Uncontrolled seizures, comorbidities and treatment side effects mean individuals are impacted in many aspects of their daily life and need a lot of support. However, support is not always available or readily accessible for affected families.
Due to the refractory nature of the condition, frequent nocturnal seizures means the risk of Sudden Unexpected Death in Epilepsy (SUDEP) is high in this patient population.

UNRAVEL...

Making change happen!

At Ring20 we are working with partners to address the unanswered research questions:

- **Diagnostics**

Ring20 are partnering with Illumina Inc who are undertaking an R&D study to see if genomic sequencing can make diagnosis easier and help us find more effective treatments.

- **Therapeutics**

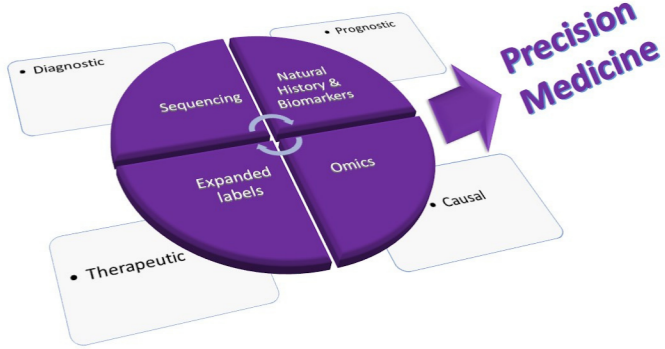
Ring20 are exploring opportunities for individuals to gain access to a wider range of treatments, but need to identify biomarkers for drug repurposing.

- **Causes**

r(20) can be diagnosed via karyotype testing or FISH. However, is the ring formation merely a symptom of an (as yet unidentified) genomic change? What effect does the 'ring' have on genes and gene expression?

- **Prognostics**

Ring20 have undertaken a Patient Reported Impact Study and are seed funding an international Natural History Study. We also have plans to start a patient registry.



Read more about our research strategy and activities on our website:
https://ring20researchsupport.co.uk/research/