Promoting research, education and continuous support to end undiagnosed and misdiagnosed Ring20 epilepsy

ring 20 research & support

@Ring20UK ring20epilepsy Ring20ResearchandSupportUK

KARYOTYPE

Diagnostic Testing Procedure for: r(20) epilepsy syndrome

We have seen the ring chromosome 20 in as few as 5% of cells, and we recommend requesting a screen for **chromosomal mosaicism**.
Since r(20) syndrome can present as a mosaic with the ring in only a small number of cells, **a minimum of 50 cells must be analyzed.**

More information:
https://ring20researchsupport.co.uk/for-medics-researchers/

Newer array technology (CGH or SNP arrays), genetic sequencing (Exome or Whole Genome), or epilepsy gene panels will NOT detect the ring chromosome and we recommend standard metaphase chromosome analysis.

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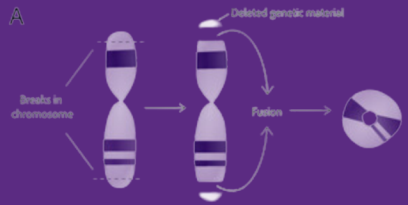
r(20) epilepsy syndrome: KNOW THE SIGNS

YOU SEE:

- Normal childhood development, *followed by*
- Sudden onset of seizures (no illness or head injury) that don't respond well to treatment
- Loss of skills
- Challenging or unusual behaviours
- Multiple seizure types, including:
  - absences (like daydreaming)
  - focal (prolonged periods of confusion) - NCSE
  - tonic clonic
- Night waking (often mistaken for night terrors)
- Seeing things that aren't there (hallucinating)

YOUR DOCTOR SEES:

- Normal MRI
- Normal genetic testing
- Abnormal background EEG (slow spike-wave complexes)



- Undiagnosed cause for epilepsy, or 'Lennox Gastaut Syndrome-like' epilepsy

Otsi says:
talk to your doctor about testing for r(20) epilepsy syndrome

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