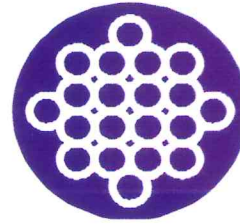


Rare Epilepsy Syndrome – Patient Support Group

ring 20
research & support uk



DOES YOUR PATIENT HAVE RING CHROMOSOME 20 SYNDROME?

R(20) syndrome can **ONLY** be identified through chromosome analysis that includes mosaic screening, or karyotype. A minimum of 50 cells must be analyzed. Newer array technology (CGH or SNP arrays) will **NOT** detect the ring chromosome. Key signs and symptoms include:

- Predominantly focal seizures
- Frequent subtle nocturnal seizures (SNS) and/or subtle nocturnal frontal lobe seizures (SNFL)
- EEG showing prolonged high voltage frontally dominant slowing intermixed with spikes or sharp waves
- EEG showing overlapping features of continuous slow spike and wave discharges in sleep (CSWS) and electrical status epilepticus in sleep (ESES)
- Normal childhood development until onset of epilepsy
- Lack of dysmorphism or other congenital malformations
- Cognitive impairment/learning difficulties/mild retardation
- Medically refractory epilepsy with no etiology identified



Contact us at:

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