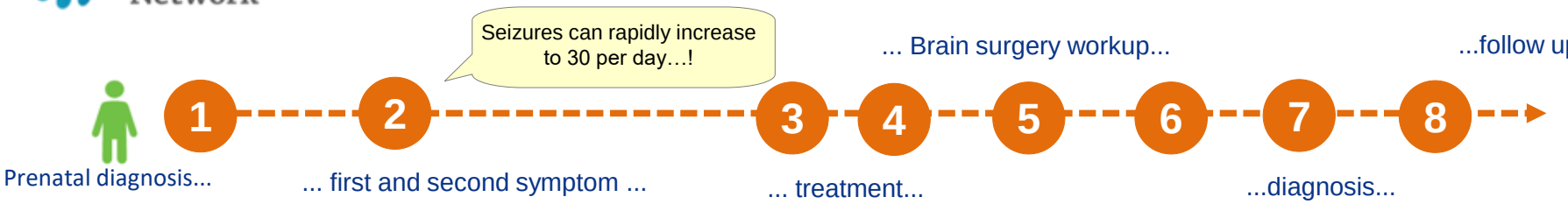
European Reference Network

Ring 20 Patient Journey



1. Prenatal diagnosis

Parent(s) diagnosed with r(20). 50% probability of having a child with r(20) – child would likely be non-mosaic (most severe form of r(20))

2. First Symptom

Major seizure, erratic behavior.
2x EEG's (private and NHS) seizures were not detected – even though they occurred during the test.

Apparently normal childhood development though presented with:
Hyperactive behavior &
hearing problems – sensitivity to loud noises.

3. Second Symtoms

Sudden and severe onset of seizures, worse at night with hallucinations.
Seizure's rapidly increasing from 5 to 30 per day. Cognitive regression.

4. Treatment

Treated for absence seizures based on parental recounts of history of episodes seen with child.

5. Treatment

Treated for focal (impaired awareness) seizures including multiple AEDs and steroids. Seizures significantly improved/abated with steroid treatment. Also treated for behavioral issues.

6. Brain surgery work-up

Referred for brain surgery workup. However, brain surgery is not a suitable treatment for r(20) patients – more works should be done to identify cause of epilepsy first.

7. Diagnosis (Age 8)

Diagnosis found by 'chance' to be r(20) – 2 yrs to diagnosis. Referred to geneticist. No recommended treatment protocol for r(20) – treatment on a trial and error basis

8. Treatment

Randomized to receive VNS therapy under clinical trial. Numerous AED's as adjunctive therapy with n benefit and range of side effects. Seizure frequency and duration gradually increasing during adolescence.

Need: Understanding of risks and consequences of having children. Listen to parent and take their concerns seriously. Family need reassurance and information about having a child with epilepsy.

Ideally: Counselling with pre-natal testing. In childhood, run tests to understand cause of apparent 'hyperactivity' and hearing issues treat.
Study EEG to diagnose seizures as a recognizable form of epilepsy.

Need: Further reassurance for patient and family. Instigate support for loss of attendance at school and cognitive regression. Need to be advise parents of risk of SUDEP and mitigating actions

Ideally: Referral to tertiary centre (paediatric neurologist). Video EEG to diagnose cause of seizures, not behavior problems or night terrors. Provide Care Plan. EHCP (or equivalent) for support at school – backed up by psychometric and neuropsychological testing. Counselling for risk of SUDEP. Advice on suitable seizure alert devices.

Need: Understand degree of behavioural problems and triggers and how to manage. And understand the seriousness and significance of brain surgery.

Ideally: Support from behaviour specialist. Identify cause for epilepsy by running genetic screening and if not identified through epilepsy panel testing or WGS, then run chromosome testing on 50-100 cells to rule out ring chromosomes prior to epilepsy surgery workup.

Note: Genetic counselling to understand the impact of the diagnosis, prognosis. Advise family of likelihood of success with polytherapy.

Ideally: Information on the diseases, genetic counselling and referral to patient support group. Explanation about level of mosaicism and what that means n terms of prognosis.
Avoid polytherapy with AED's.

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9. Follow Up (Age 11)

Genetic testing as part of genetic research study in USA concluded no loss of DNA – ring was simply formed by ends of chromosome 'sticking together'.

10. Follow Up (Age 15)

Parents requested to try ketogenic dietary therapy, though David taken off after 3-weeks due to suspected MCAD (6 months later non-founded through skin biopsy screening).

Seizure control was adversely impacted, and family left without support of KDT team or paed neuro.

5. Follow Up (Age 17-19)

Parents requested to try ketogenic dietary therapy again – but had to be referred to centre that could manage child in transition.

KDT improved seizure control (50%), alertness and mood.

On 4th VNS battery – every operation carries increased risk of infection due to reopening of old wound/scare tissue

12. Follow Up (Age 21)

Changed from Carbamazepine to Oxcarbazepine due to reduced longer term side effects (osteoporosis)

Other AED's have been removed.

Seizures gradually increasing in frequency and intensity with age currently 4-6 per day, lasting 20-30mins each (frequent episodes of NCSE)

Also tonic Clonic seizures over last few years.

13. Life-long Follow Up

Need: Support to understand how to implement KDT and workload, potential improvements (potential cognition, mood and alertness as well as seizure improvement) as well as challenges .

Ideally: Screen for suitability for KDT before prescribing .

Ideally: Ensure KDT is implemented with appropriate dietetic support and monitoring to check both ketone and glucose levels

Note: Support in further education and readiness for workplace. Understand prognosis? Lack of research (and no clinical trials in r(20)) mean that there is no information on the natural history of the disease, though experience thus far suggests seizures worsen over time. Applications for Support Dog unsuccessful due to continuous changes in treatment – need stability for 6 months to be eligible. Opportunities for employment low due to health and safety risk and lack of support in the workplace. Applications for benefits challenging (as with many living with uncontrolled with epilepsy). Basic needs misunderstood. Would not be able to live independently without support.

Ideally: Regular reviews with paed neuro/neurologist to monitor change in condition. Regular blood tests to check for side effects of long-term or changes in AED's.