Why Aren't They Listening?

Acknowledging the challenges facing individuals with rare and complex epilepsies and their families in the UK

UK Rare Epilepsies Together

Aim

UK Rare Epilepsies Together (UKRET) are looking to identify the **key shared challenges** and **perceived gaps in care and support** for individuals with **rare and complex epilepsies** in the UK. By spotlighting these challenges, we can now explore practical solutions and define recommendations for integration into the UK healthcare system. Our goal is to improve quality of life (QoL) outcomes for these individuals and reduce the impact on families, whilst optimising valuable healthcare professionals' (HCP) time and experience.

Scope

Rare and complex epilepsies are typically conditions where seizures are refractory to treatment, therapy and/or surgery and are associated with many co-morbidities. The challenges commonly faced by children and adults living with these epilepsies and their families remain underacknowledged in current literature and medical practice.

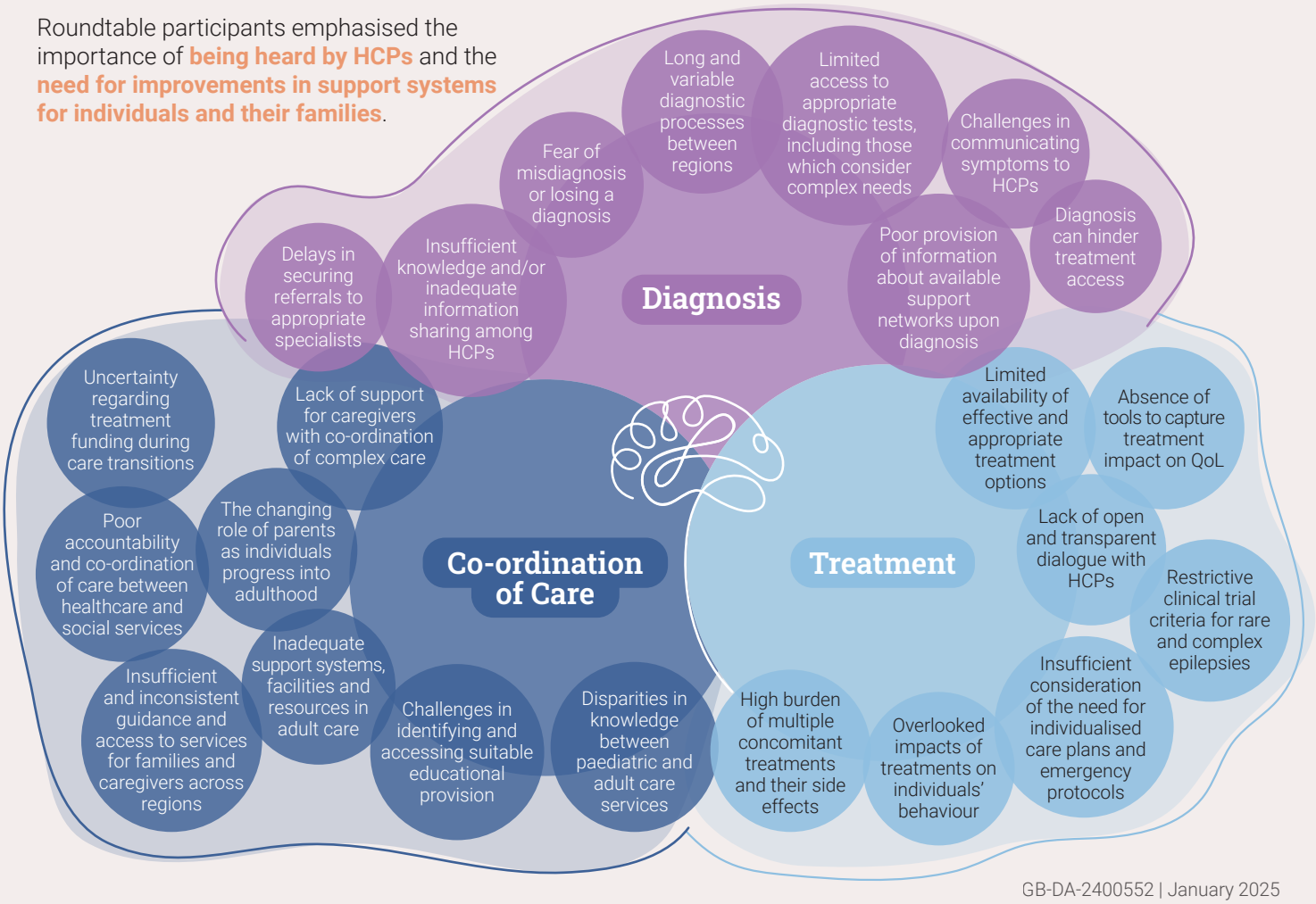
By working together as the UKRET network, we amplify our voice to drive change for the often-overlooked rare and complex epilepsy community, whose needs differ from those with controlled epilepsies.

Methods

- Targeted literature searches identified materials reporting on challenges faced by individuals with rare epilepsies and their families.
- These materials shaped the agenda for a roundtable meeting on the 27th September 2024.
  - Nine leaders of rare epilepsy patient groups and caregivers themselves, **shared their lived experiences and those of individuals in their communities**.
  - Discussions covered challenges with **diagnosis**, **treatment** and **co-ordination of care**.
- Findings were validated by other UKRET network members who were unable to attend the meeting.

Challenges

Roundtable participants emphasised the importance of **being heard by HCPs** and the **need for improvements in support systems for individuals and their families**.



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Why Does Overcoming These Challenges Matter?

Diagnosis

The process of getting a diagnosis for a rare and complex epilepsy can be difficult and stressful for individuals and their families. Those affected may need to push for referrals and may have to repeatedly explain their symptoms to different HCPs, **leaving them feeling ignored and overwhelmed. This is tough and unfair for those without the means or skills to advocate for themselves**. While some might quickly receive an initial diagnosis of epilepsy, finding the root cause often takes years due to a lack of awareness about more rare and complex conditions, **leading to frustration and uncertainty**.

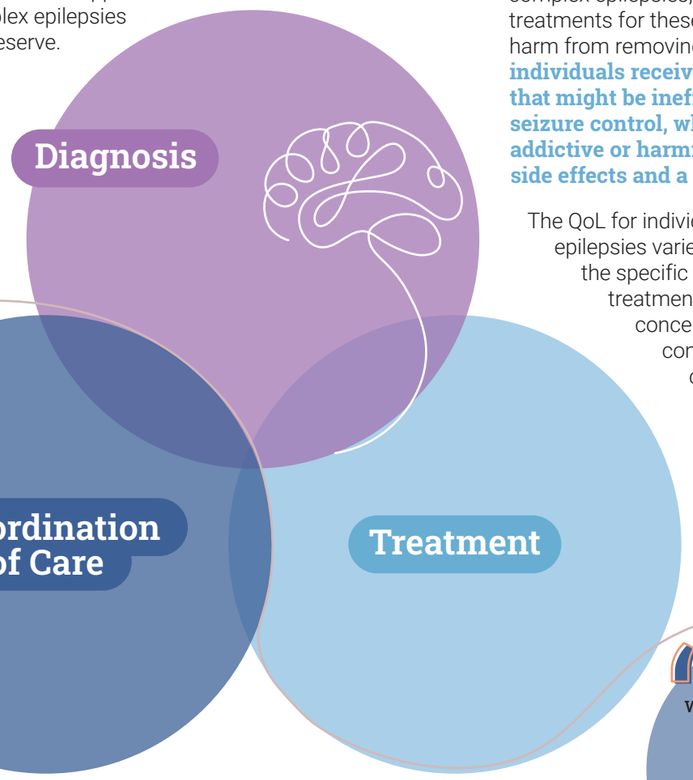
Even with recent advancements, genetic testing doesn't always provide answers. HCPs often rely heavily on this form of testing, potentially overlooking other diagnostic methods (e.g. immunological, metabolic and chromosomal testing), and may fail to seek advice and share learnings. **This prolongs the diagnostic odyssey, preventing early access to available treatments and support networks that individuals and families come to rely upon**.

Additionally, misdiagnoses are common and **can lead to less effective treatments being prescribed and more confusion and stress**. This highlights the urgent need for better diagnostic practices and comprehensive support to ensure individuals with rare and complex epilepsies and their families receive the help they deserve.

> "There's real variability in ease of getting a diagnosis – it often comes down to regional postcode lottery"

> "Diagnostic tests are not the final barrier, and even though we know it's epilepsy and they're being treated for epilepsy, the test doesn't always give you the diagnosis you're searching for"

> "I repeatedly hear of the struggle of being heard by HCPs"



Co-ordination of Care

Transitioning to adult care can reveal major gaps in support for individuals with rare and complex epilepsies and their families. For adults, the focus shifts to purely seizure management and access to a multidisciplinary team disappears. Non-pharmacological treatments such as ketogenic dietary therapy are not well provisioned, often overlooked or inappropriately discontinued in older children. Combined with the possibility of expensive treatments being stopped, this **limits available treatments and hinders care**. There are few epilepsy specialists in adult care, so individuals typically need to see their general practitioner who knows little about their condition.

Regardless of age, there is inadequate support with co-ordination of care and limited access to care co-ordinators, forcing individuals and families to handle all appointments and referrals independently. **This responsibility increases the burden of care on families**, who must juggle these tasks whilst caring for their loved one(s). Families often encounter poor guidance and information, even on important topics like care plans, emergency protocols and Sudden Unexpected Death in Epilepsy (SUDEP). This is likely due in part to a shortage of epilepsy specialist nurses (ESNs), limited use of SUDEP and seizure safety tools and a confusing healthcare system. These factors collectively **increase the risks that can endanger the lives of individuals with these epilepsies.**

Navigating paperwork and financing for broader social support (examples below), **can be daunting for families who are already overwhelmed**.

- Access to a social worker
- National Health Service Continuing Healthcare (NHS CHC)
- Education and healthcare plans
- Power of attorney or deputyship
- Benefits:
  - Disability Living Allowance
  - Personal Independence Payment
  - Universal Credit

Added challenges include the lack of referrals for individuals and families to support services such as counselling, mental health care and intrafamilial genetic testing. **As a result, families may not be accessing all the support available**.

> "HCPs often don't remove medications from a treatment regimen, they just keep adding to it"

> "Trust is essential; it's challenging for doctors to openly say they don't know"

> "The individual voiced that he wished to come off the drug but imagine how terrible it must be for kids who can't voice how they're feeling"

> "There's no single 'solution' treatment that will work for the condition, even for two family members with the same condition"

Treatment

With a lack of treatment guidelines for rare and complex epilepsies, HCPs often continue to add treatments for these individuals, fearing potential harm from removing existing ones. **This leads to individuals receiving numerous treatments that might be ineffective in improving seizure control, whilst being potentially addictive or harmful, resulting in serious side effects and a reduced QoL**.

The QoL for individuals with rare and complex epilepsies varies significantly depending on the specific condition. Monitoring of treatment effectiveness typically concentrates solely on seizure control, neglecting other co-morbidities. While trying to manage seizures is crucial, HCPs neglect to consider factors such as impact of treatments on overall wellbeing, behaviour and sleep. Families often attempt to convey these needs but feel overlooked by HCPs, **resulting in individuals not receiving holistic care and management**.

Due to the wide spectrum of rare and complex epilepsies, treatments that benefit one person may not be effective for another. HCPs adopt a trial-and-error approach to treatment, but often fail to clearly communicate to individuals and their families the impact of this process or how diagnosis and funding may limit available treatments. **This lack of transparency may lead to mistrust, leaving families feeling isolated and unempowered**.

There is a shortage of appropriate tools to assess treatment effectiveness and impact on QoL, as well as a lack of clinical trials for individuals with rare and complex epilepsies. **This leaves individuals and families feeling stuck with limited options and support**.

> "Maintaining support through the NHS CHC scheme can be challenging and may require legal support to challenge eligibility decisions"

> "The presence of a social worker is crucial, but not all families have access to one"

> "Individuals with rare epilepsies are living to adulthood, and so we need people interested in them and all their care needs"

> "As soon as you fall over the cliff into adult neurology, they primarily focus on seizure control, and trying to access a multidisciplinary team is a lost cause"

> "I would like to stay [in paediatric care] for as long as I can honestly"

> "Often 1.5 ESNs are assigned to 460 epilepsy patients whereas nurses in paediatric oncology have 45 patients on their caseload. ESNs should be available to all individuals with epilepsies equitably, people with rare epilepsies will need more interaction with ESNs due to changes in treatments and requiring access to more servic"

What's Next?

UKRET plan to host a multistakeholder roundtable in **early 2025** to explore **practical solutions** to the identified challenges. Insights from both roundtable meetings will be compiled into a report detailing the challenges facing individuals with rare and complex epilepsies, potential solutions and our proposed recommendations.

Contact Details

If you have any questions or are interested in learning more, please contact Allison Watson via allison@ukret.com.

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Abbreviations

ESNs: epilepsy specialist nurses; **HCPs:** healthcare professionals; **NHS CHC:** National Health Service Continuing Healthcare; **QoL:** quality of life; **SUDEP:** Sudden Unexpected Death in Epilepsy; **UKRET:** UK Rare Epilepsies Together; **UK:** United Kingdom.

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