Poster #P464

Barriers to and benefits of identifying patients with developmental and epileptic encephalopathies in adult care settings

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Overview

QUESTION

What are the barriers to, and benefits of identifying developmental and epileptic encephalopathy (DEE) in adults within residential care settings?

INVESTIGATION

A qualitative study using Interpretative Phenomenological Analysis (IPA) explored the experiences of caregivers and HCPs across the UK, Germany, France, and Spain. Interviews focused on diagnostic barriers, referral behaviours, and opportunities to improve care for adults with suspected or confirmed DEE.

RESULTS

Variations in care provision

Specialist centres were equipped with multidisciplinary teams, regular patient reviews, and staff trained in complex epilepsy care, which enabled earlier diagnosis and holistic management.

In contrast, non-specialist settings often lacked DEE awareness, structured review processes, and confidence to escalate for genetic testing.

Recognising a patient with DEE

Subtle cognitive or behavioural changes were key indicators, but identification depended on frequent, structured assessments and staff familiarity with DEE.

Regular reviews and detailed observation notes helped flag patients for diagnostic re-evaluation.

Barriers to diagnosis

Delays were linked to unfamiliarity with DEE, limited access to EEG or genetic testing, and attitudinal barriers such as reluctance to disrupt stable medication regimes.

Inconsistent input from MDTs and a lack of family advocacy also hindered timely referrals.

Benefits of a diagnosis

A confirmed DEE diagnosis enabled tailored treatment, greater access to specialist services, and improved alertness and communication in patients.

It also reduced emergency interventions and provided clarity and emotional reassurance for families and care teams.

CONCLUSIONS

Improving DEE identification in adult care may be supported by regular developmental reviews, greater awareness among non-specialist teams, and broader outcome tracking beyond seizure control. Early and accurate diagnosis can lead to better quality of life through more appropriate treatment, increased independence, and greater emotional and cognitive wellbeing.

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Background

Despite significant advances in the diagnosis of developmental and epileptic encephalopathy (DEE) in childhood, many adults in long-term residential or daycare settings may remain undiagnosed. Often labelled as having "generalized epilepsy with additional needs," these individuals may not have been reviewed in light of evolving genetic and diagnostic criteria. This has profound implications not only for clinical care and medication management but, importantly, for overall quality of life (QoL).

Objective

This qualitative study seeks to explore the experiences of adults with and without a formal DEE diagnosis in adult residential care. It aims to uncover the barriers to diagnosis in these settings, the factors that can unlock a diagnostic review, understand the specific care challenges and missed opportunities, and highlight the potential benefits of accurate, timely diagnosis.

Methods

DESIGN

This was a qualitative, exploratory study using Interpretative Phenomenological Analysis to uncover the experiences of adults with DEE in residential care.

PARTICIPANTS

Caregivers and healthcare professionals (HCPs) from the UK, Germany, France, and Spain were interviewed, representing a range of roles including specialist nurses, psychologists, and family members.

APPROACH

Semi-structured, 60-minute interviews were conducted via Zoom or in person. A phased approach allowed for early insight generation in the UK, followed by theme development across mainland Europe. An idiographic lens was used to identify themes and regional variations, informing understanding of unmet needs in care.

Results

The study included 22 participants (8 caregivers, 8 specialist nurses, 6 other HCPs) from 4 countries and varied care settings, including specialist and non-specialist environments.

VARIATIONS IN CARE PROVISION

Specialist centres were identified as well-equipped to support diagnostic review for potential DEE:

- Typically have strong multidisciplinary teams (MDTs) including therapists, nurses, psychologists, and physicians.
- Offer balanced management of seizures, behaviour, cognition, and emotional wellbeing.
- Regular reviews include nurses' insights from daily care, highlighting subtle changes.

"As nurses we attend reviews. We are with the patients 24/7, so have a much better idea of behaviours and emotional development."
– Specialist nurse, France

Non-specialist settings could improve support for DEE diagnosis by:

- Increasing DEE awareness and providing basic training for staff.
- Implementing simple structured tools to monitor cognitive and emotional changes.
- Facilitating regular reviews to capture broader patient progress.

"Staff won't be aware of the DEE group of conditions and therefore do not push for a diagnosis rethink."
– Specialist nurse, UK

Table 1. Demographics

Respondent	UK	Spain	Germany	France	TOTAL
Parents/ family members of those with DEE in adult care	2	2	2	2	8
Specialist nurses working with those with DEE in adult care	3	2	1	2	8
Other HCPs working with those with DEE in adult care	1 nurse in residential care; 1 hospital nurse in epilepsy clinic	1 psychologist	1 GP linked to specialist centre	1 nurse manager; 1 central medical coordinator	6
TOTAL	7	5	4	6	22

DEE, developmental and epileptic encephalopathy; GP, general practitioner; HCP, healthcare professional.

RECOGNISING A PATIENT WITH DEE

Clinical reviews often prioritise seizure management, but expanding the scope to developmental assessments supports better identification of patients who may benefit from a diagnostic review:

- More frequent reviews can promptly highlight behavioural or cognitive changes.
- Structured prompts or tools help identify developmental regressions or progress.
- Broadening outcome tracking to include cognitive and emotional milestones promotes holistic patient care.

"Residents here are reviewed almost weekly – it means we can observe and quickly note changes."
– Nurse coordinator, France

BARRIERS TO DIAGNOSIS

Family dynamics and patient preferences affect diagnostic reassessment:

- Patients may choose not to undergo further testing.
- Families can be reluctant to revisit diagnoses due to exhaustion or fear of destabilisation after potentially experiencing a lifetime of drug adjustments.
- In some cases, especially with older patients, there may be no family members to advocate for them.

Care team structure and experience influence diagnosis:

- MDTs lacking consistent input may miss diagnostic cues.
- Adult care teams often have less exposure to rare epilepsy syndromes.

Attitudinal barriers include:

- Clinician reluctance to challenge stable seizure control or medication routines.
- Caution around altering established treatments.

"'More or less controlled' is not enough, we need to be providing the specific aspects to control quality of life."
– Nurse, Spain

Process constraints include:

- Limited consultation time focused predominantly on medication.
- Difficulties accessing timely diagnostic tests (EEG, MRI, genetics).

BENEFITS OF A DIAGNOSIS

For patients, improved QoL:

- Better access to appropriate medication and therapies enhances autonomy and engagement.
- Improved alertness, communication, motor skills, and personality expression.
- Tailored support from specialists (e.g. speech, physical, occupational therapists)

For families, clarity and relief:

- Reduces emotional burden through understanding the condition's origins.
- Enables realistic expectations and present-focused QoL improvements.

"The diagnosis, when we got it, was a shock in one respect and in another respect, it was a huge relief for us."
– Parent, Germany

For clinical teams, targeted care:

- Allows tailored interventions and staff training.
- May reduce unnecessary emergency interventions.

For health systems, efficiency gains:

- Streamlined care reduces hospitalisation and emergency calls.
- Optimises resource use and facilitates access to specialist services.

"Funding authorities want evidence of the trajectory of a condition, and this is easier to provide with a definitive diagnosis."
– Specialist Nurse, UK

PATIENT STORY

Two 30-year-old brothers – one with a Dravet syndrome diagnosis, one without

The specialist nurse was involved in a case of two brothers in their 30s. One had a Dravet syndrome diagnosis and lived in a nursing home where they were unable to deliver specialist care.

The parents wanted him to remain local, so did not want to push for him to be cared for in one of the specialist centres. His younger brother had shown signs of similar symptoms and patterns but did not have a Dravet syndrome diagnosis.

The nursing staff had observed his cognitive and behavioural symptoms and pushed for a referral for genetic testing, but that took 2 years. Unfortunately, he died of sudden unexpected death in epilepsy (SUDEP) during that period. When writing the report for his death certificate, they questioned whether more prompt genetic testing could have led to a more effective management plan and potential prevention of his death by SUDEP.

PATIENT STORY

A 65-year-old patient who has been put forward for genetic testing following a change in GP

A 65-year-old patient was experiencing 45 seizures on a monthly basis. He was frequently in hospital and discharged on the maximum dose of medication. He has been on strong medication for a very long time – but the specialist nurse felt that he might do better on fewer medications, that he might be more alert and at a lower risk of SUDEP. Fewer seizures would mean more mobility, more connected to the activities of the residential home, and feeling more comfortable in general.

The specialist nurse was familiar with Dravet syndrome and other rare syndromic epilepsies, so she referred him to the GP for a local review. He had limited family to intervene, so it was down to the nurse to advocate for him. The GP responded by saying that there was no change with this patient, so the medication regime will need to stay the same, and he would not refer him to a consultant.

This response led to a switch in GP, who has reviewed the patient and put him forward for genetic testing.

Conclusions

- This study highlights the barriers to diagnosing DEEs in adults within residential and daycare settings, particularly where care is focused narrowly on seizure management.
- A broader view that includes cognitive, behavioural, and emotional development can support earlier recognition and more appropriate intervention.
- Raising awareness of DEE characteristics and the diverse benefits of diagnosis across patients, families, and care systems may lead to more timely diagnoses and better QoL.
- With greater confidence and knowledge, non-specialist teams can play a key role in identifying DEEs and achieving more tailored, effective care.

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