Improving the developmental and epileptic encephalopathy diagnostic process: Recommendations to address the challenges

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Background

- Developmental and epileptic encephalopathies (DEEs) encompass a group of severe diseases, typically beginning in early childhood.[1,2]
- DEEs are characterised not only by frequent and generally pharmacoresistant seizures but also by developmental impairment, in which both the underlying aetiology and the epileptic activity itself contribute to the neurodevelopmental disability.[1,2]
- Delayed diagnosis of DEEs contributes to suboptimal treatment, which may further worsen prognosis; therefore, improving diagnostic efficiency is a clinical priority globally.[1–3]

Objective

- To develop practical recommendations and considerations to support early diagnosis of DEEs by analysing the diagnostic process for DEEs worldwide.

Methods

- Paediatric and adult neurologists with expertise in DEEs took part in an international Expert Input Forum in August 2025.
- Participants independently identified diagnostic barriers, as well as associated concerns, which were discussed in workshops to prioritise cross-regional challenges and opportunity.
- Thematic analysis was applied to derive globally relevant recommendations/considerations.

Results

IDENTIFIED CHALLENGES

- Participants (N=11) from Australia, Belgium, Brazil, Hong Kong, Israel, Mexico, Taiwan and Türkiye prioritised 11 challenges **(Table 1)**.
- The identified challenges fell into three themes:

1. **Diagnostic and aetiological complexity** – encompassing the fundamental difficulties in accurately identifying DEEs, distinguishing them from other conditions, understanding their genetic and metabolic underpinnings and predicting their trajectory.
2. **Gaps in comprehensive management and care standardisation** – focusing on challenges in providing appropriate and early treatment access and managing non-seizure symptoms, as well as the lack of consistent, evidence-based protocols.
3. **Patient and care pathway continuity challenges** – focusing on issues related to effective communication with caregivers and ensuring continuity of care across different life stages.

Table 1 Prioritised challenges (N=11) in early diagnosis of DEEs and associated themes

Diagnostic and aetiological complexity (n=5)	Gaps in comprehensive management and care standardisation (n=4)	Patient and care pathway continuity challenges (n=2)
Recognition and referral to specialised epilepsy centres by primary care physicians (e.g. GPs and paediatricians) and general neurologists	**Poorly defined non-seizure symptoms and lack of assessment** (limited time in consultations)	**Communication to caregivers** to educate on and manage expectations
Difficulties in differentiating between DEEs and other disorders	**Lack of evidence or guidance for co-management of non-seizure symptoms**	**Transition between paediatric and adult care** and DEE diagnosis among adults
Unpredictability of outcomes (correlation between genetics and phenotype)	**Lack of guidelines/ screening/protocols**	
Lack of understanding of genetics, including testing and interpretation, and **lack of standardisation**	**Lack of access to appropriate early treatment**	
Lack of consideration of metabolic syndromes		

DEE, developmental and epileptic encephalopathy; GP, general practitioner.

RECOMMENDATIONS AND CONSIDERATIONS

- Based on the identified challenges, the experts proposed a number of short- and long-term actionable recommendations/considerations **(Table 2)**.
- Short-term recommendations/considerations included broad education to improve early recognition of DEEs and facilitate aetiology-specific diagnosis with genetic testing, education on transition to adult care and additional support for caregivers.
- Long-term recommendations/considerations included increased research on disease outcomes, impact of early treatment interventions and ways to manage non-seizure symptoms, as well as data generation to facilitate development of evidence-based guidelines and screening tools.

Table 2 Actionable recommendations and considerations to address identified challenges associated with early diagnosis of DEEs

Short-term actionable recommendations/considerations	Long-term actionable recommendations/considerations
Educational resources for GPs and paediatricians in both primary care and neurology are needed to **improve early recognition and referral**	**Advanced research and amplified data generation** would help to mitigate the **unpredictability of disease outcomes**
Resources for caregivers should be developed to **communicate effectively** and **manage expectations** about the diagnosis and prognosis	A **multidisciplinary approach and more research are needed to define non-seizure symptoms**; the standard of care should include the assessment of non-seizure symptoms and QoL, with the development of **disease-/syndrome-specific QoL assessments**
Dissemination of the ILAE definitions and classifications of epilepsy syndromes would **increase knowledge/understanding of DEEs** and facilitate differentiation from other disorders	**Clear scientific evidence** is needed for the **development of guidelines, protocols and screening tools**
Training of adult neurologists is essential to **support the transition from paediatric to adult care** and DEE diagnosis among adults	**Research** is needed to understand the **benefits and harms of early interventional treatment**
Training on genetics for neurologists, paediatricians, primary healthcare workers and medical students would **improve and standardise understanding of genetics,** including testing and interpretation	
Education is needed to support the **inclusion of metabolic screening** in standard diagnostic work-up	

DEE, developmental and epileptic encephalopathy; GP, general practitioner; ILAE, International League Against Epilepsy; QoL, quality of life.

Conclusions

The recommendations and considerations developed by this international Expert Input Forum underscore the urgent need for expanded research, enhanced education and structured diagnostic work-up to improve and accelerate DEE diagnostic pathways globally and optimise the management of patients and their caregivers.

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