

Patient Journeys: A Tool to Raise Awareness on the Evolution, Common Needs and Critical Issues of Patients with Rare and Complex Epilepsies



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Introduction

The heterogeneous and complex nature of the rare and complex epilepsies often results in sub-optimal care. Over 130 rare forms of epilepsy have to date been identified across all ages and aetiologies, and that number is growing. These rare epilepsies may be genetic, structural, immune, infectious, metabolic, syndromic. Some are well-managed by anti-convulsant medication, some are surgically treatable, some have no current effective treatment. While seizures are the common hallmark, each disease has debilitating co-morbidities affecting neurodevelopment, quality of life and mortality. ERN EpiCARE's objectives are to improve diagnosis, treatment and awareness of rare and complex epilepsies, but the breadth and variety of the patient cohort creates significant challenges in meeting these objectives.

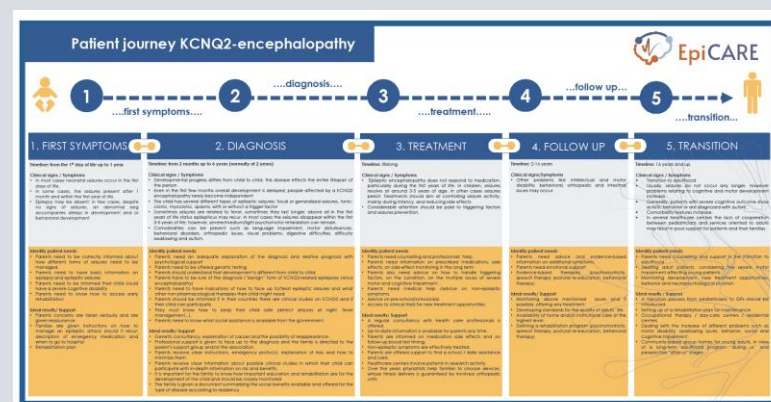
Methodology



consensual experiences of patients with rare diseases

Iterative workshops

- ✓ Identify Gaps and area for improvement
- ✓ summarize steps of care pathways
- ✓ List elements considered positively by those affected
- ✓ highlight common needs and relevant differences



Results

Several commonalities emerged - even where etiologies and seizure types and age of onset differed. Lifelong challenges and gaps in care were common to all. While seizure control or cessation was key across all the mapped epilepsies, it was universally highlighted that this was only one aspect of syndrome management. Notably, every Patient Journey author emphasized that the specific epilepsy that they had narrated was a complex syndrome with often devastating co-morbidities: life with a rare and complex epilepsy is a marathon for patients and those who care for them. The ePAG mapped the common unmet needs identified through the Patient Journey comparison and developed a position paper which they presented at EpiCARE's 2020 AGM. EpiCARE clinicians and researchers were invited to consider and address the full spectrum of needs of this patient community; ePAG counselled the adoption of a multi-disciplinary approach from the point of diagnosis, and continuing throughout the patient's lifetime.

The following key priorities were highlighted:

1. Seizure control is a primary goal. Specialist centres are best placed to achieve this:- through sharing expertise and experience and developing common prescribing guidelines and surgical pathways.
2. Seizure control is impossible without early, accurate diagnosis. Specialist centres are well-placed to work with the ePAG to better educate first instance clinicians such as neurologists and pediatricians in syndromes, seizure types, etiologies and progression. We invited the development and dissemination of diagnostic protocols.
3. Family support: targeted support is central to help the whole family cope with the patient's complex, life-long needs and achieve the best possible quality of life. Discussion re SUDEP should be tackled straightforwardly and early.
4. Diverse co-morbidities require a multidisciplinary approach and cross-specialty cooperation. Timely, consistent access to therapies is crucial for child development and future quality of life. There should be regular review throughout patient life. Psychiatric and psychological support are vital from early childhood and throughout adulthood.
5. Transition to adult services was universally described - across countries and across syndromes - as 'like falling off a cliff'. This must be improved, and adult neurologists must be better educated in treating so-called 'childhood' epilepsies. At the point of transition there should be comprehensive liaison between treating clinicians and adult social care to support independent/semi-independent living, tertiary education and assisted work opportunities and socialization within the patient's local community.

Outcomes

When developing its five-year plan for 2022-2027, with ePAG input EpiCARE established several new Working Groups (WGs) and Special Interest Groups (SIGs). These include WGs on "Epilepsy Beyond Seizures," "Transition," "Guidelines" and on supporting the ePAG. SIGs include "Pre-surgical Evaluation Guidelines," "Surgical Guidelines," and "Diagnostic Gaps". Each of these working groups has ePAG representation.

EpiCARE clinicians also worked with the ePAG to oversee and approve the production of information leaflets for individual syndromes which describe best practice for clinicians and which set out the key features in patient-centric language for the patient and caregiver.

The Patient Journeys, together with the leaflets, are available on the EpiCARE website, and at EpiCARE treatment centres. Clinicians and patients alike are directed to them to give a quick and accurate overview of the symptoms, treatment options and progression of these rare and ultra-rare syndromes that most first instance clinicians will come across perhaps just once or twice in the course of their practice.

Available Patient Journeys: [Glut1 Deficiency Syndrome](#), [Hypothalamic Hamartoma Syndrome](#), [Dravet Syndrome](#), [Alternating Hemiplegia of Childhood](#), [KCNQ2](#)

About to be published : [Lennox Gastaut Syndrome](#), [Ring Chromosome 20 Syndrome](#), [CDKL5](#)

Conclusion

From these exchanges it became clear that ERN EpiCARE needed to widen its focus beyond seizure control and consider the holistic needs of this patient community. Notably, every patient advocate emphasised that the specific rare epilepsy that they represented was a complex syndrome that required a multi-disciplinary approach starting from the point of diagnosis and continuing throughout the patient's lifetime. After the ePAG presentation of the Patient Journeys in one of EpiCARE's annual meetings and the subsequent position paper, ERN EpiCARE agreed to develop and disseminate standards of care based on the identified common unmet needs under a clinical pathway for all patients with a rare and complex epilepsy.

References

<https://epi-care.eu/about-epilepsies/>

<https://epi-care.eu/patient-and-caregiver-leaflets/>

The Rare and Complex Epilepsies Common Unmet Needs within the Patient Community, poster, ECRD 2020

Acknowledgement

Thanks for collaboration Gregori Cabanach, Emma Nott, Vedrana Bibic, Irena Bibic and all ePAG patient representatives and the community who collaborated on the draft contents and translations.