

# The Rare and Complex Epilepsies

## Common Unmet Needs within the Patient Community



### Introduction

Rare and complex epilepsies are numerous, diverse and have many, and differing, aetiologies. The heterogeneous and sometimes subtle nature of their clinical presentation and the huge variation in first symptoms often result in a delay in securing a timely diagnosis. For many sufferers there are few or no available treatment options. While seizures are the common hallmark of all rare and complex epilepsies, each syndrome has co-morbidities that are often equally, or more, debilitating.

ERN EpiCARE has been set up to improve the prognosis and quality of lives for the children affected. But with such a large and varied patient cohort, how can the Network best understand and meet their complex and diverse needs?

### Methodology

Patient representatives within the EpiCARE ePAG each detailed the individual needs of their children in a syndrome specific 'Patient Journey'. This documented the natural history of that particular rare syndrome, which they presented to the group in a workshop. As each advocate gained greater insight into the differing aetiologies, complexities, symptoms and prognoses of other rare and complex epilepsies, a number of commonalities emerged - even where aetiologies and seizure types were very different. Advocates then mapped the common needs affecting patients and families across all syndromes and used these to develop a position paper presented for discussion at ERN EpiCARE's Annual General Meeting.

### Results

Seizure control/cessation is the key for each epileptic syndrome. However, it is only one aspect of syndrome management and ERN EpiCARE must widen its focus beyond seizure control and consider the holistic needs of this patient community. Notably, every Patient Advocate emphasized that the specific rare epilepsy that they represented was a complex syndrome that requires 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 and its consequent 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.**

### Summary

Life with a rare and complex epilepsy is a marathon for patients and for those who love and care for them. This should be openly recognized at the point of diagnosis, when the parents need and deserve competent and comprehensive guidance on all aspects of managing syndromes and maintaining the best possible quality of life. Early diagnosis and family-centred communication are the cornerstones of optimizing the family's ability to cope with the complex and usually life-long needs of the patient. Doctors should consider and address the patient's lifelong increased susceptibility to mental illness. More medical understanding of SUDEP - mechanism, risk factors, effective preventative measures - is overdue, as is the provision of detailed and careful information to patients/families about this.

### Common unmet needs highlighted as key priority are:

#### 1. Early, correct diagnosis

This will require better diagnostic protocols and more frontline training in seizure types, particularly the more subtle types. If seizures are suspected or the patient presents with suspected autism, unexplained 'tics' or developmental delays, epilepsy should be considered as a potential cause. Specialist epilepsy centres should be considered even where MRI scans, CAT scans and EEGs appear normal.

#### 2. Seizure control

Send patients to specialist centres early; develop common prescribing guidelines and surgical pathways; share expertise and experience.

#### 3. Informed families

The tension between seizure control and quality of life should be acknowledged, with side effects from treatment outlined and explained. Being well-informed and supported is central to the family's ability to cope with the patient's complex, life-long needs. There should be discussion surrounding SUDEP.

#### 4. Multidisciplinary Care

Timely access to therapies, equipment and education is crucial to the child's current and future quality of life and independence. Diverse and often catastrophic co-morbidities require cross-specialty cooperation. Input from clinical psychologists at a very early stage should be standard: with a multi-disciplinary medical team, along with parents, driving the child's education plan.

#### 5. Regular Review

Even where seizures are controlled through surgery or medication, the MDT should keep the patient under review. Ongoing psychiatric and psychological assessment and support is vital, with the current transition from paediatric to adult services universally described - across syndromes and across countries - as 'like falling off a cliff.'

#### 6. Rehabilitation

For the young adult, a common unmet need is an adequate rehabilitation programme to support independent/semi-independent living and life opportunities - tertiary education, socialisation within the local community, assisted work opportunities.