



# Beyond the physical – The true impact of rare disease



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- co-lead for EPAG EpiCARE ERN for rare and complex epilepsies

EJPRD General Assembly: Measuring the burden of rare diseases

14<sup>th</sup> September 2021



# About Ring20 Research and Support UK CIO

- Only patient support group for r(20) in the world
- Currently support > 120 families worldwide
- Signpost to information/connect families
- Raising funds for research



# ring 20

Research & support uk





European Patient Advocacy Group



EpiCARE

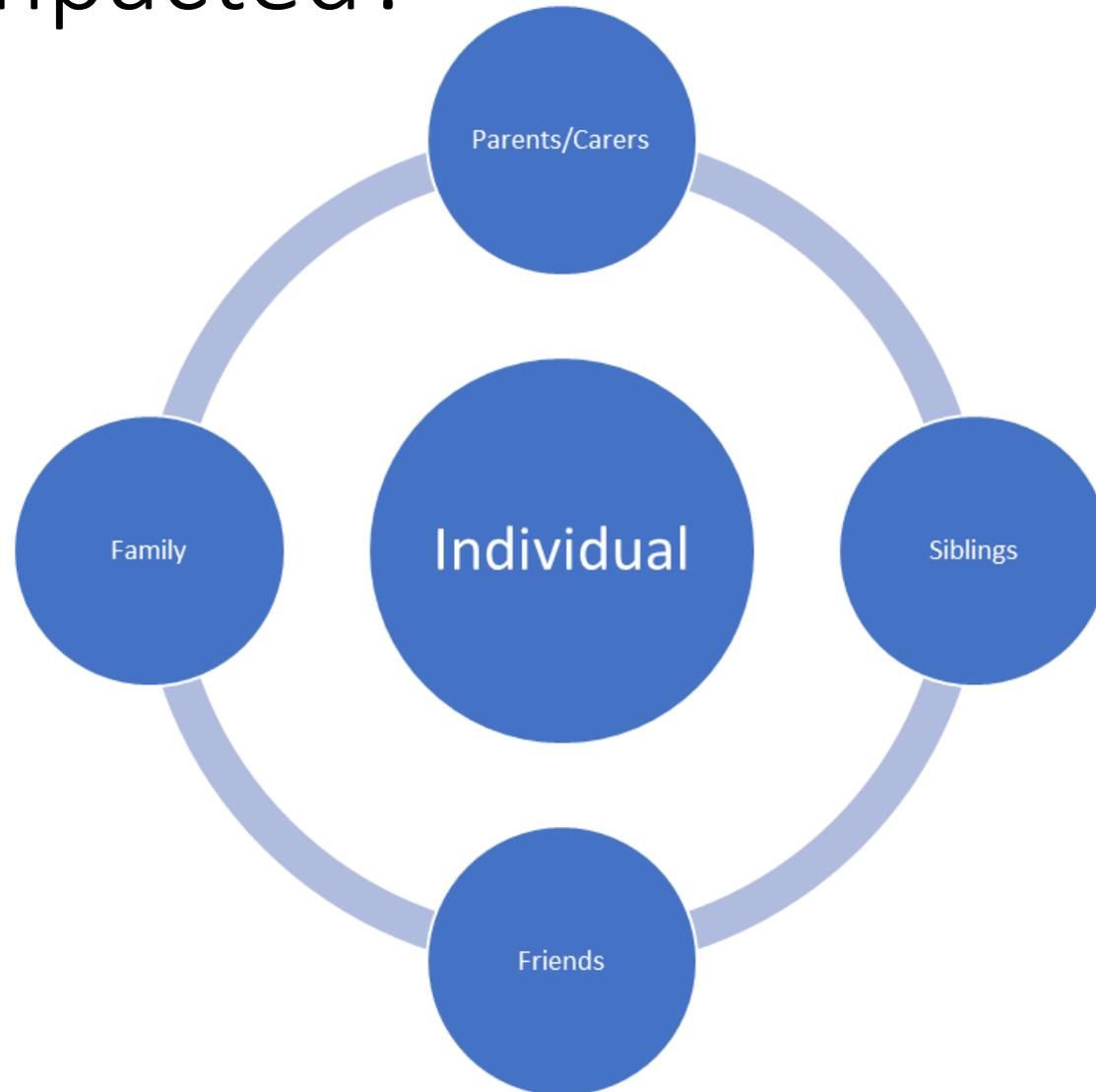
# EpiCARE ERN



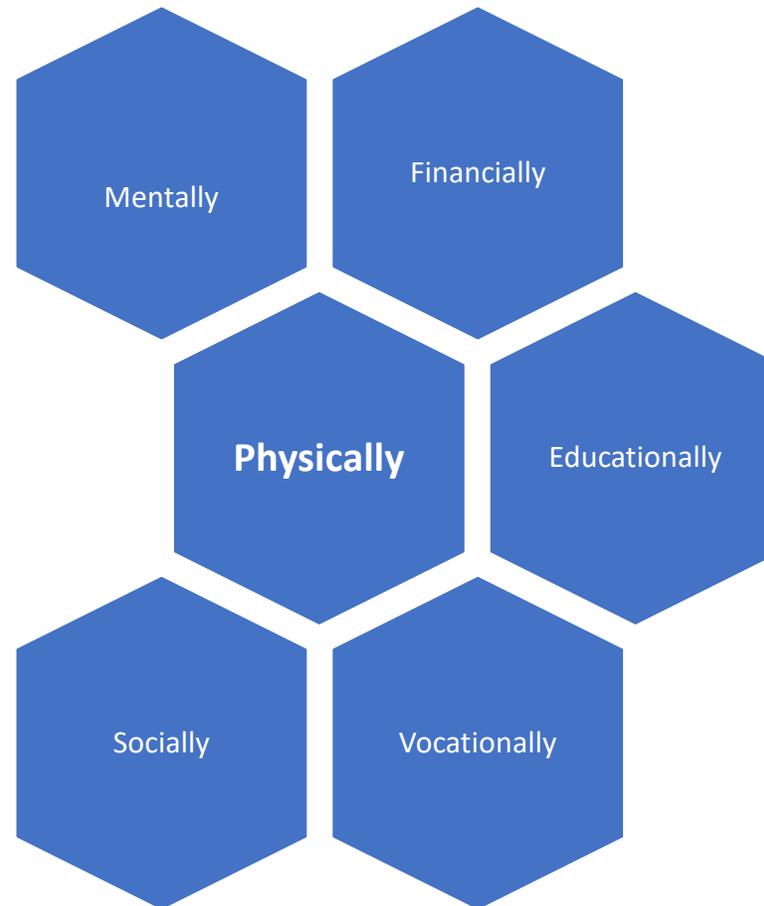
# The burden of rare disease



# Who is impacted?



# How are they impacted?



# When are they impacted?

First symptoms

Diagnosis

Childhood

Transition

Adulthood



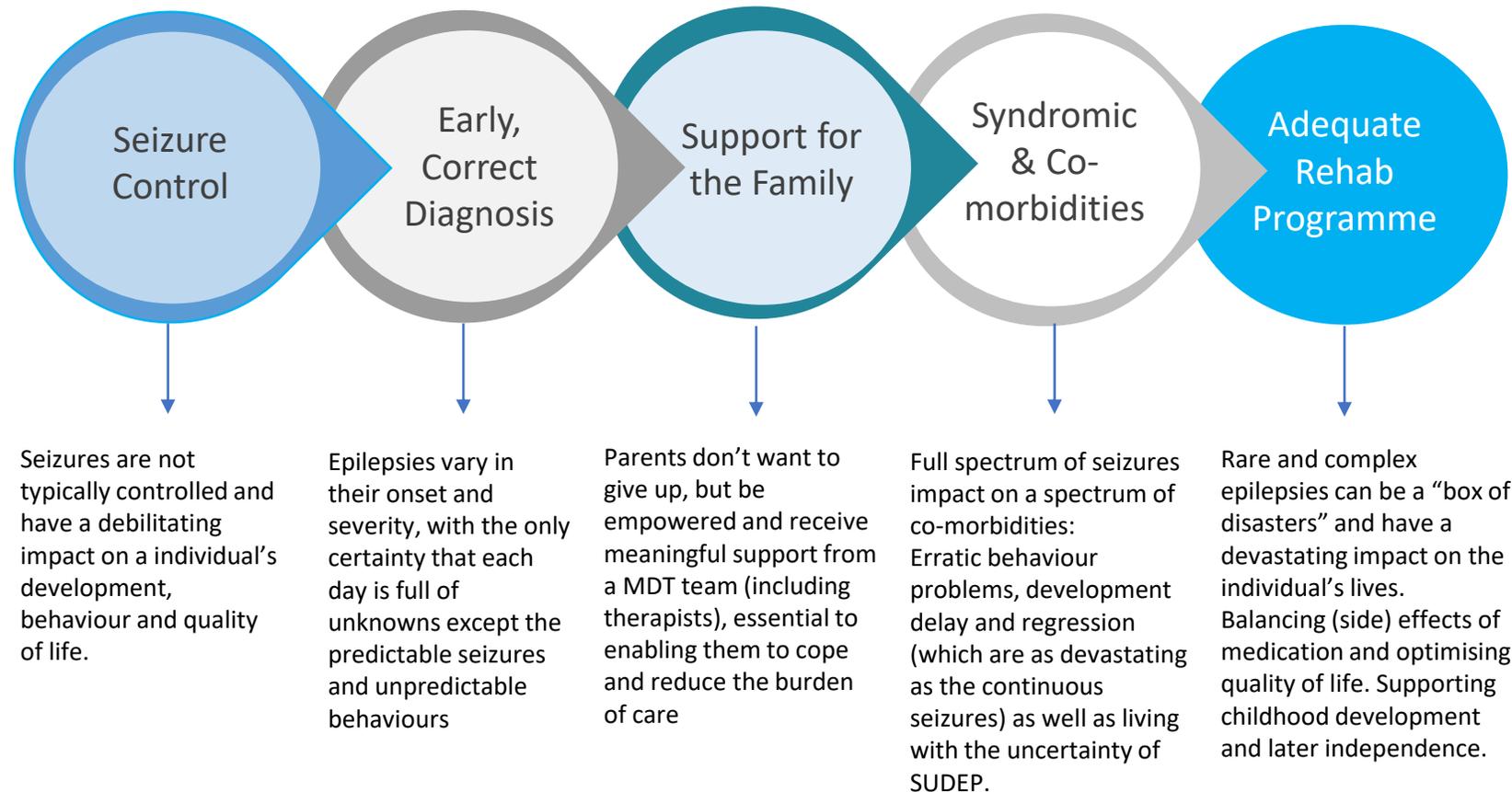
# Evidence: Patient Journeys



**The Rare and Complex Epilepsies  
Common Unmet Needs Within the Patient Community  
P088 Theme 3**

# Our Aims, Your Aims: Quality care throughout life to enable children and families to lead healthy lives!

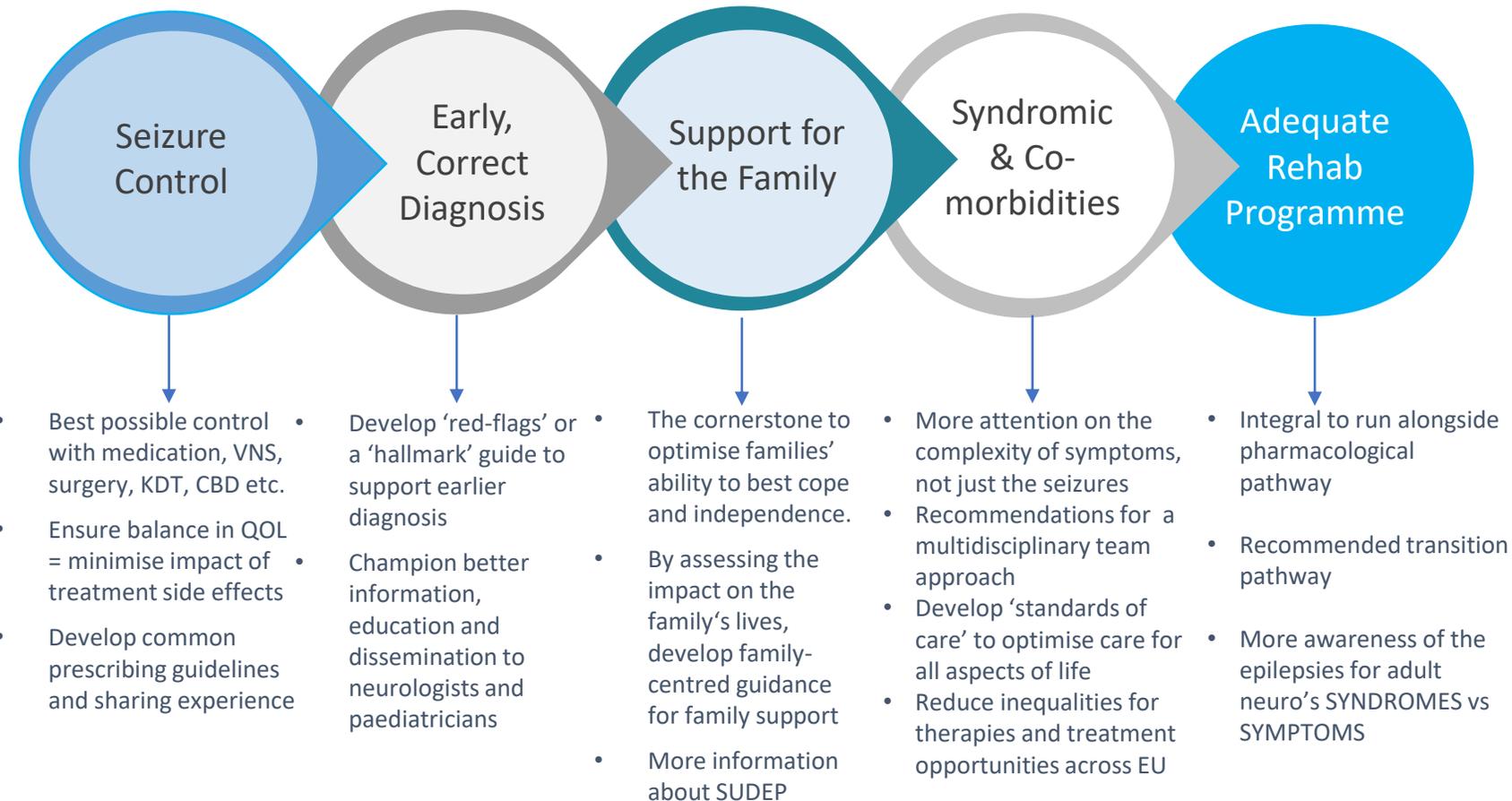
Seizure control is the key for many Rare & Complex Epilepsy syndromes however it is only one aspect of the individual and ERN EpiCARE needs to go beyond seizure control and think about the full spectrum of needs of this patient community



common unmet needs

# Our Aims, Your Aims: Quality care throughout life to enable children and families to lead healthy lives!

Seizure control is the key for many Rare & Complex Epilepsy syndromes however it is only one aspect of the individual and ERN EpiCARE needs to go beyond seizure control and think about the full spectrum of needs of this patient community



recommendations

# What is needed?

- **Information**

- Available
- Understandable
- Timely

- **Support**

- Heard/Believed
- Valued
- Connected

Stakeholders:

- Policymakers
- Workplaces
- Social workers
- Healthcare professionals
- Payers
- Regulators
- Society
- Schools
- Pharma/Industry

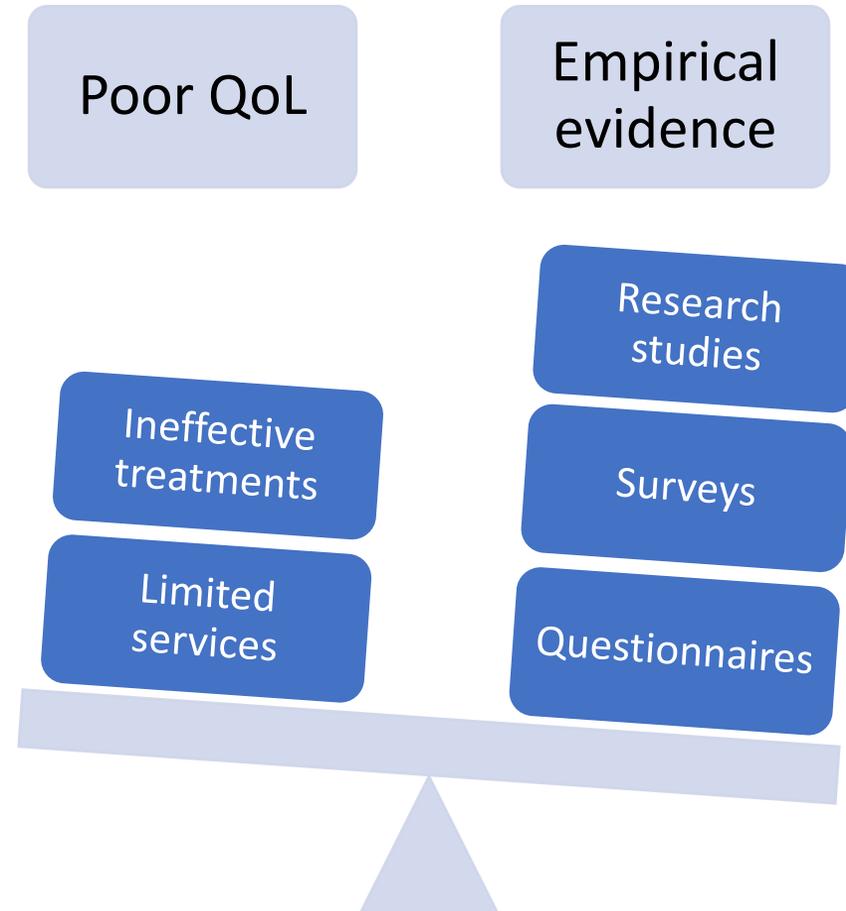
# Measuring burden...



Not all rare diseases are equal

How can you measure 6,000+ RDs effectively?

# What is not needed?



# Why is it important?



When medicine and science  
don't have all the  
answers...YET!

# RECOGNITION OF RD's IS KEY!



Call for all stakeholders  
to work together and  
**ACT NOW!**  
To optimise QoL



# Questions?



For more information or if you can help:

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website: [www.ring20researchsupport.co.uk](http://www.ring20researchsupport.co.uk)

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