

# Beyond the physical – The true impact of rare disease



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

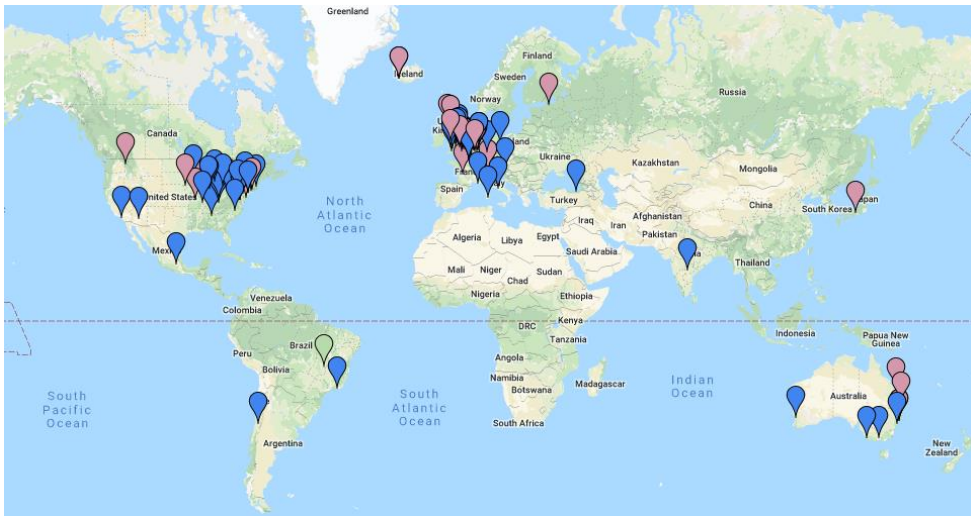
EJPRD General Assembly: Measuring the burden of rare diseases

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

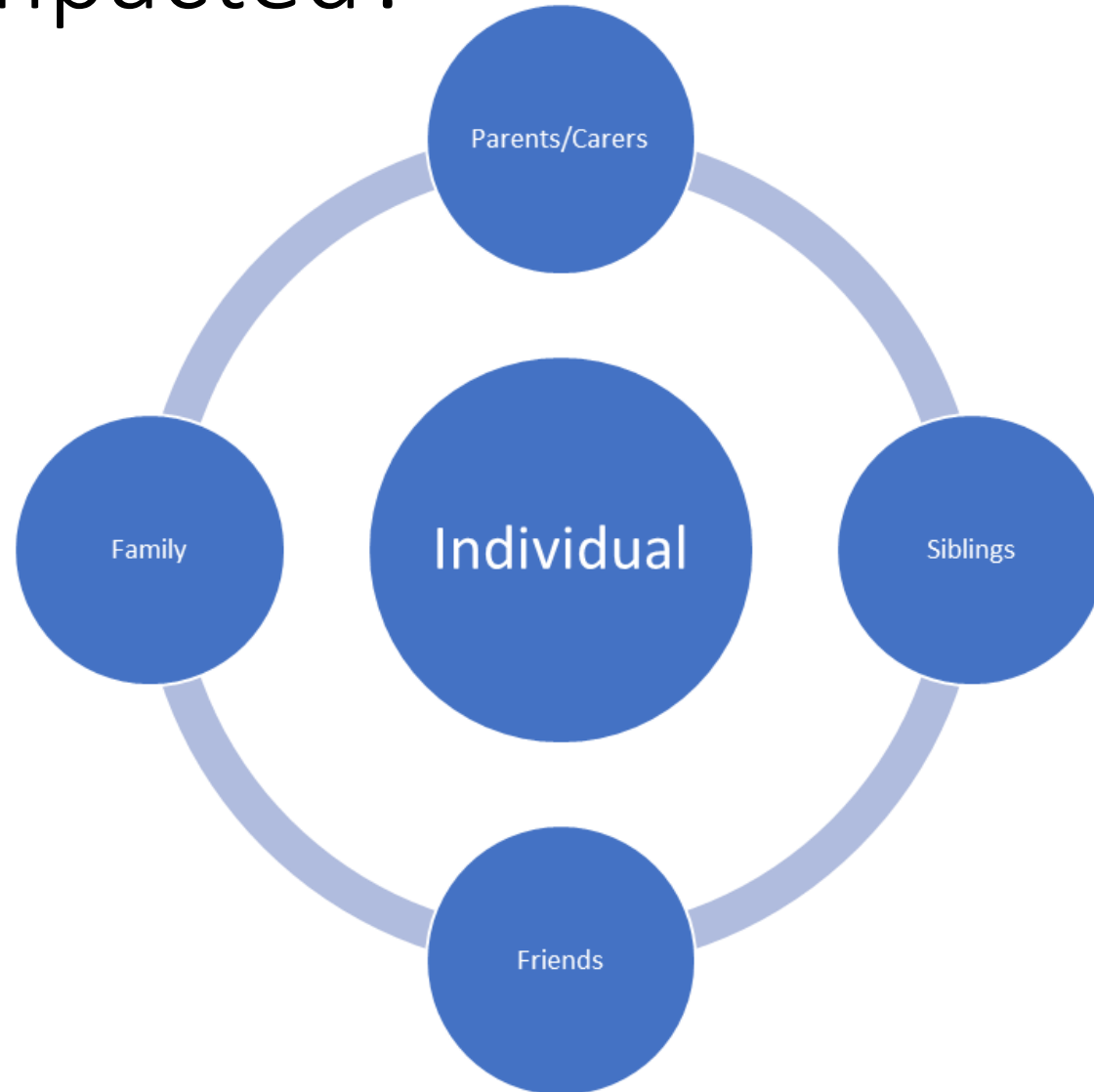




# The burden of rare disease

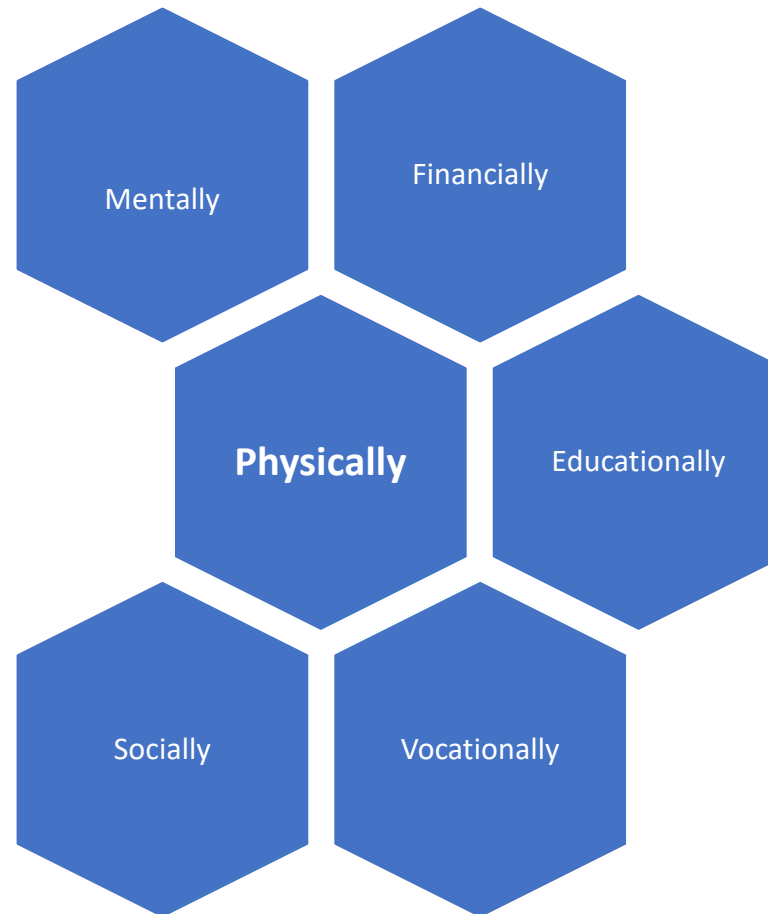


# Who is impacted?





# How are they impacted?



# When are they impacted?

First symptoms

Diagnosis

Childhood

Transition

Adulthood

# Evidence: Patient Journeys

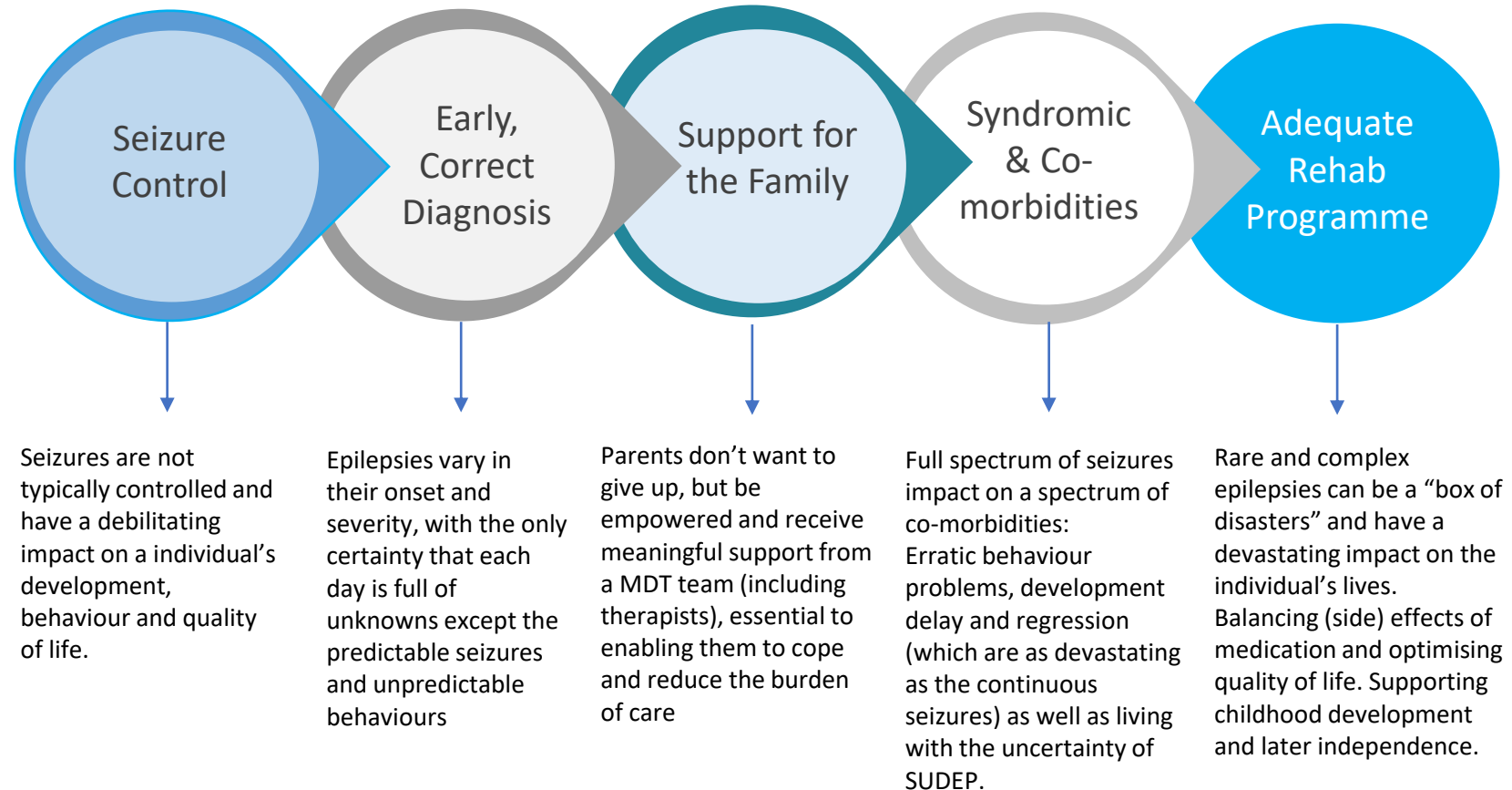


**The Rare and Complex Epilepsies  
Common Unmet Needs Within the Patient Community  
P088 Theme 3**



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

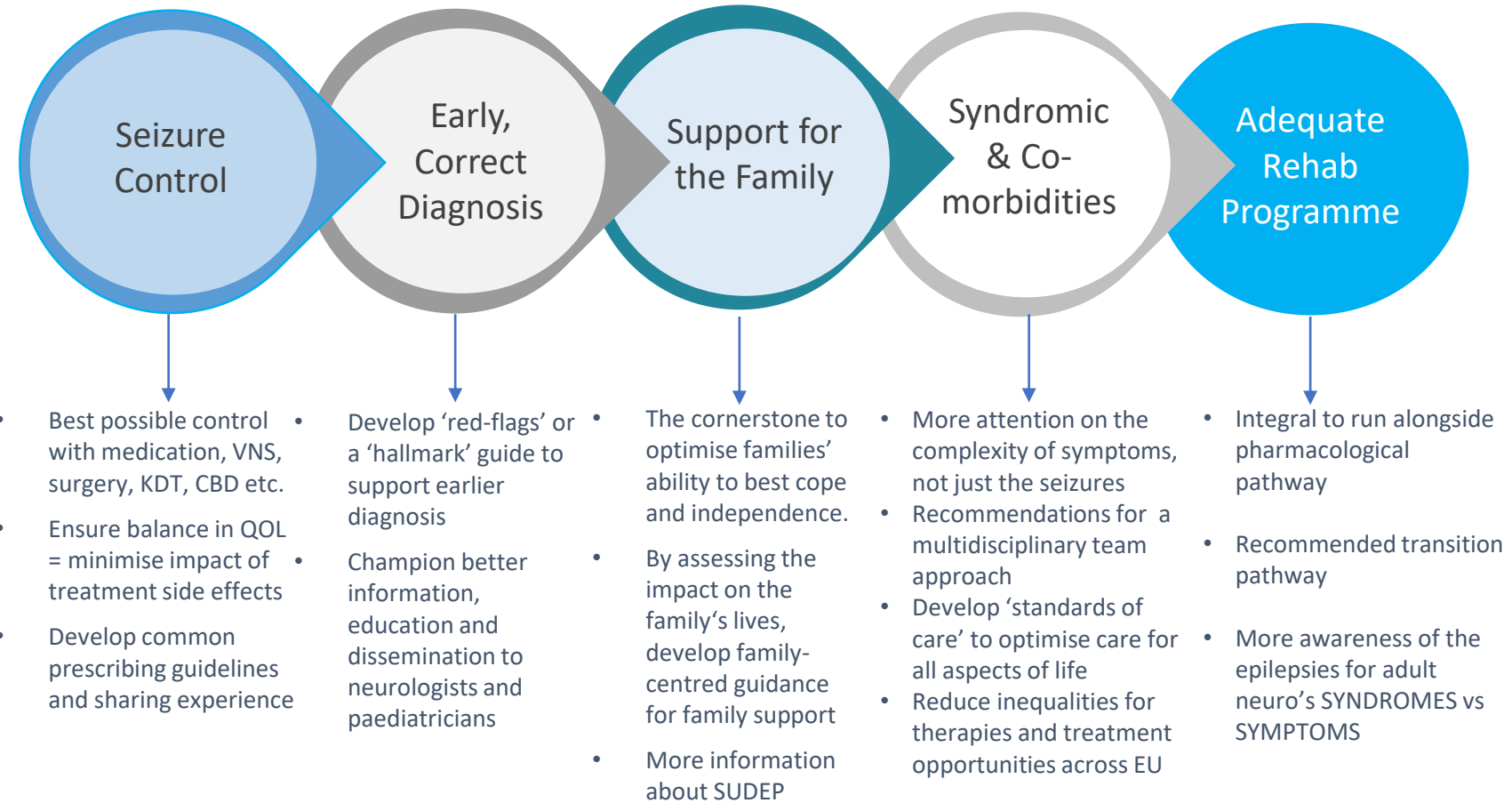
Seizure control is the key for many Rare & Complex Epilepsy syndromes however it is only one aspect of the individual and ERN EpiCARE needs to go beyond seizure control and think about the full spectrum of needs of this patient community



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

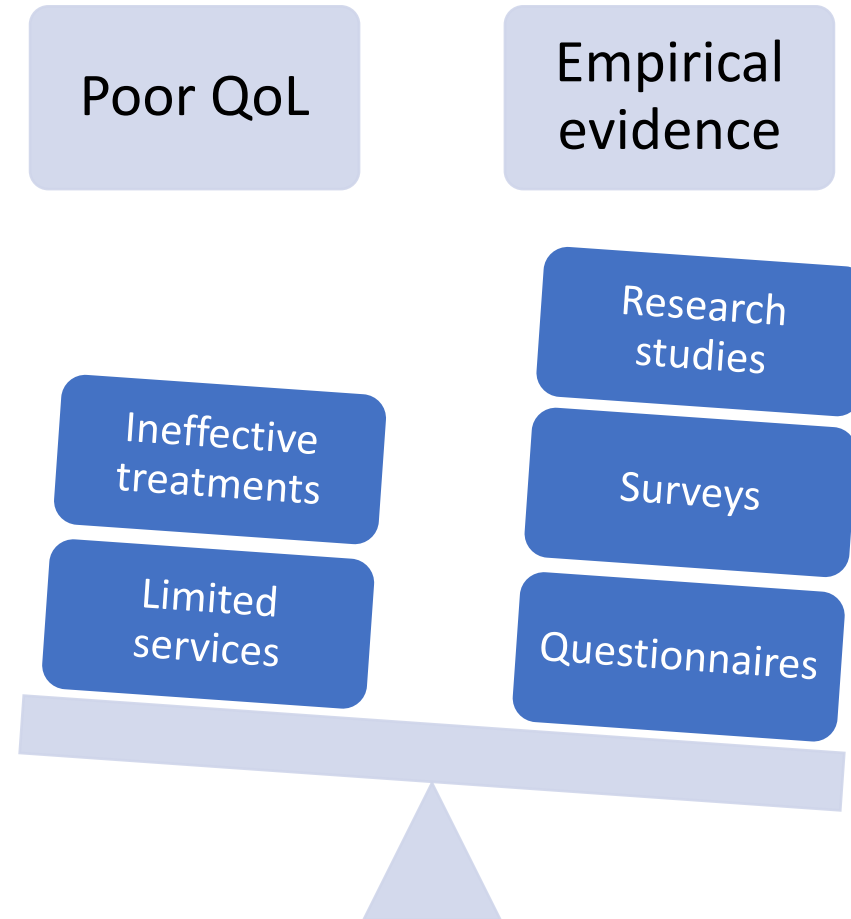


A word cloud of various stakeholders in a handwritten blue font, arranged in a diamond shape. The central and largest text is 'Healthcare professionals'. Other words include 'Policymakers', 'Workplaces', 'Social workers', 'Payers', 'Regulators', 'Society', 'Schools', and 'Pharma/Industry'.

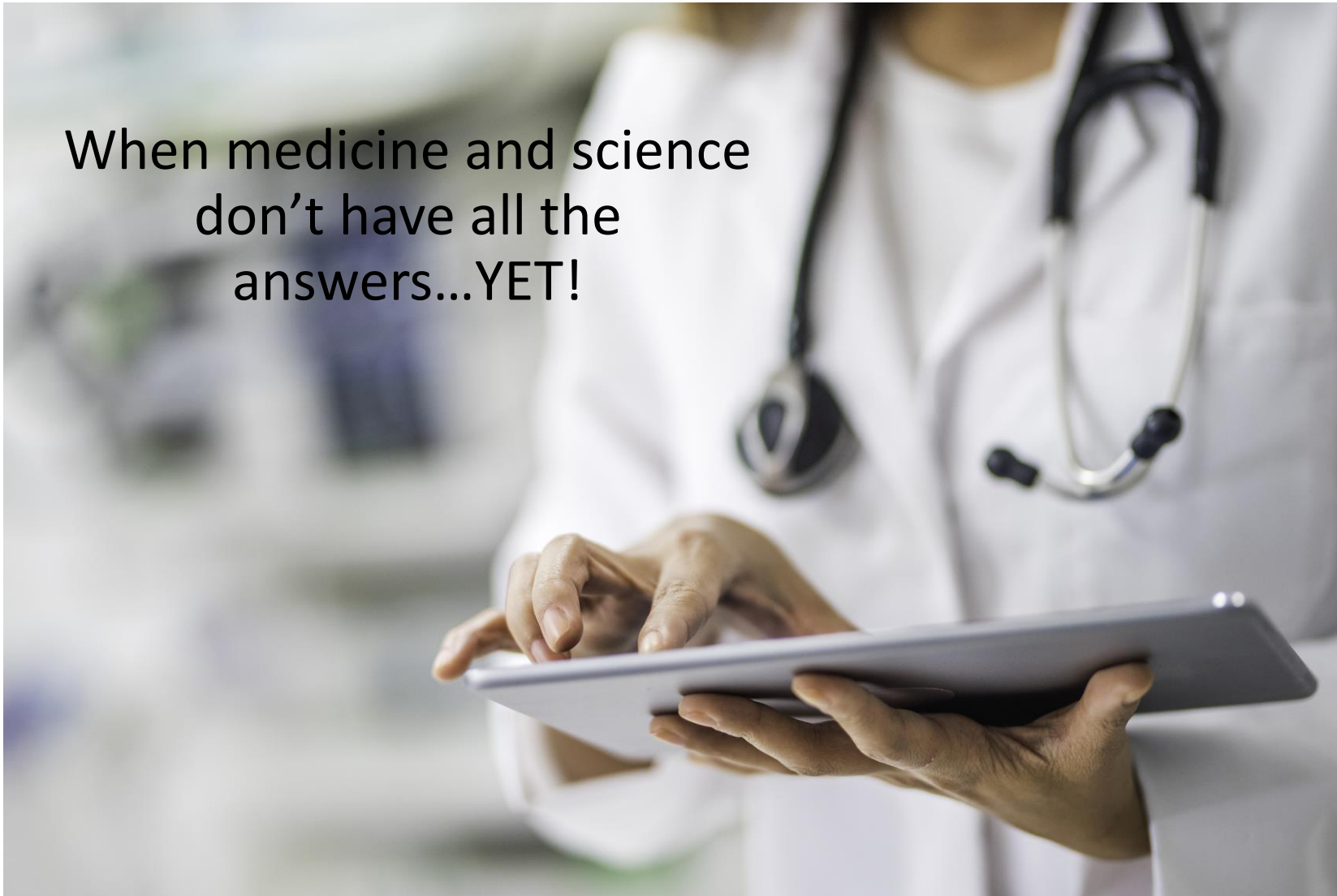
# Measuring burden...



# What is not needed?




# Why is it important?





# 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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