Challenges relating to research and innovation in the area of rare diseases

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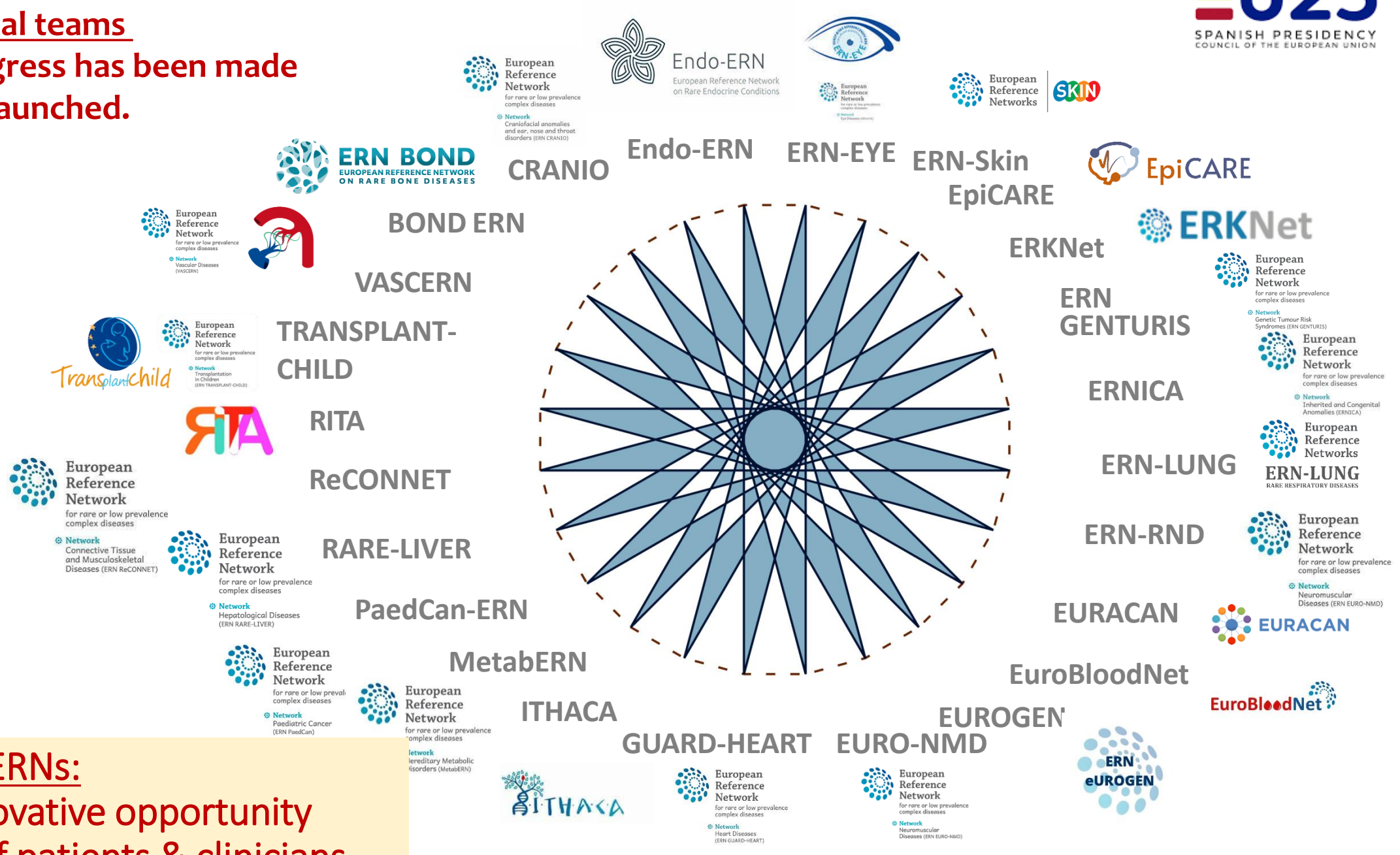
Fostering Research is one of the core areas of work of the ERN medical teams & important progress has been made since they were launched.



Creation of 24 ERNs:
A unique & innovative opportunity at the service of patients & clinicians

Courtesy ERN EYE

15TH CONGRESS OF THE
EUROPEAN PAEDIATRIC NEUROLOGY SOCIETY
20-24 JUNE 2023
PRAGUE
CZECH REPUBLIC

EPNS 2023
European Paediatric Neurology Society Congress

SPANISH PRESIDENCY
COUNCIL OF THE EUROPEAN UNION

RARE NEUROLOGICAL DISORDERS IN CHILDREN AND THE ROLE OF THE

European Reference Networks

Thursday, 22 June 2023
17.15-18.30PM

Alexis Arzimanoglou
Coordinator of EpiCARE (European Reference Network on Rare and Complex Epilepsies)

Maurizio Scarpa
Coordinator of MetabERN (European Reference Network on Hereditary Metabolic Disorders)

Juan Darío Ortigoza-Escobar
Paediatric Neurologist at SJD Barcelona Children's Hospital

Alain Verloes
Coordinator of ERN ITHACA (European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders)

Teresinha Evangelista
Coordinator of EURO-NMD (European Reference Network on Neuromuscular Diseases)

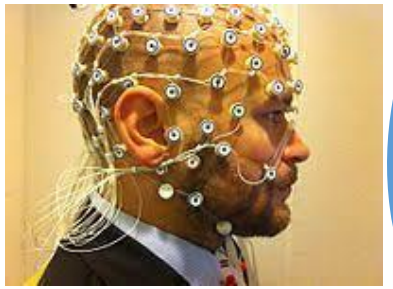
Concerted actions to motivate next-generation clinicians to be interested in rare diseases,

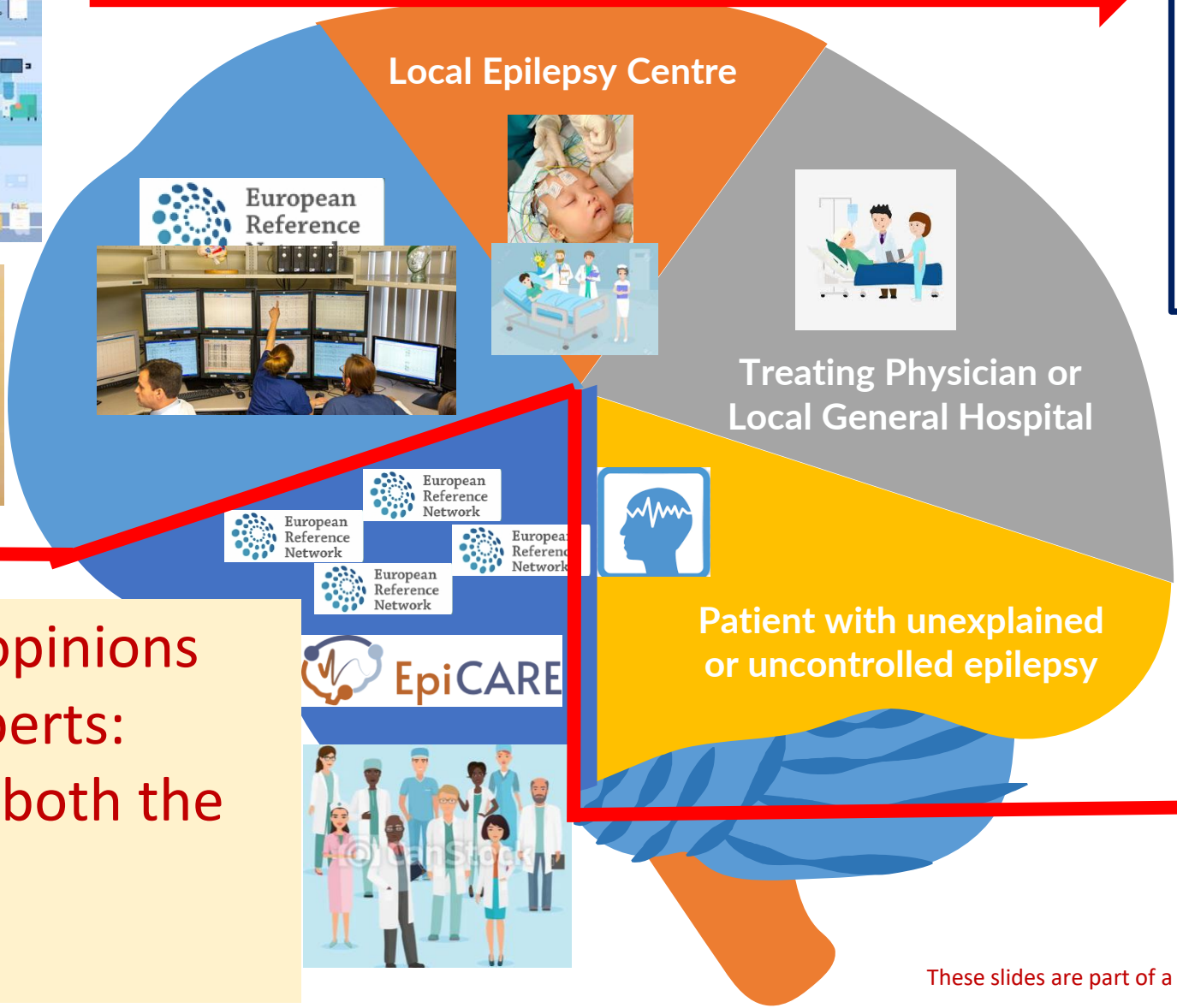
also sharing highly interactive activities ***between ultra-experts***

were rare before the creation of the ERNs.
They became common practice in less than 6 years !

EpiCARE

Care Pathways: Before we know if a patient has a Rare or Complex disease … he/she already has a disease





National level (proximity) centres with expertise are the cornerstone.

ERN to share opinions with other experts: reassuring for both the patients & the physicians

Take Home Messages - 1

1. The ERNs are **NOT** virtual.

They represent a vast consortium of EU based experts, shaping collaborations at national and international level, together with patient advocates, aiming to understand the complex mechanisms of hundreds of rare diseases.

De facto they also **shape the national health care systems** for early diagnosis of both common and rare diseases, the open the pathways for the discovery of better treatments.

A given disease can be considered rare only once diagnosed

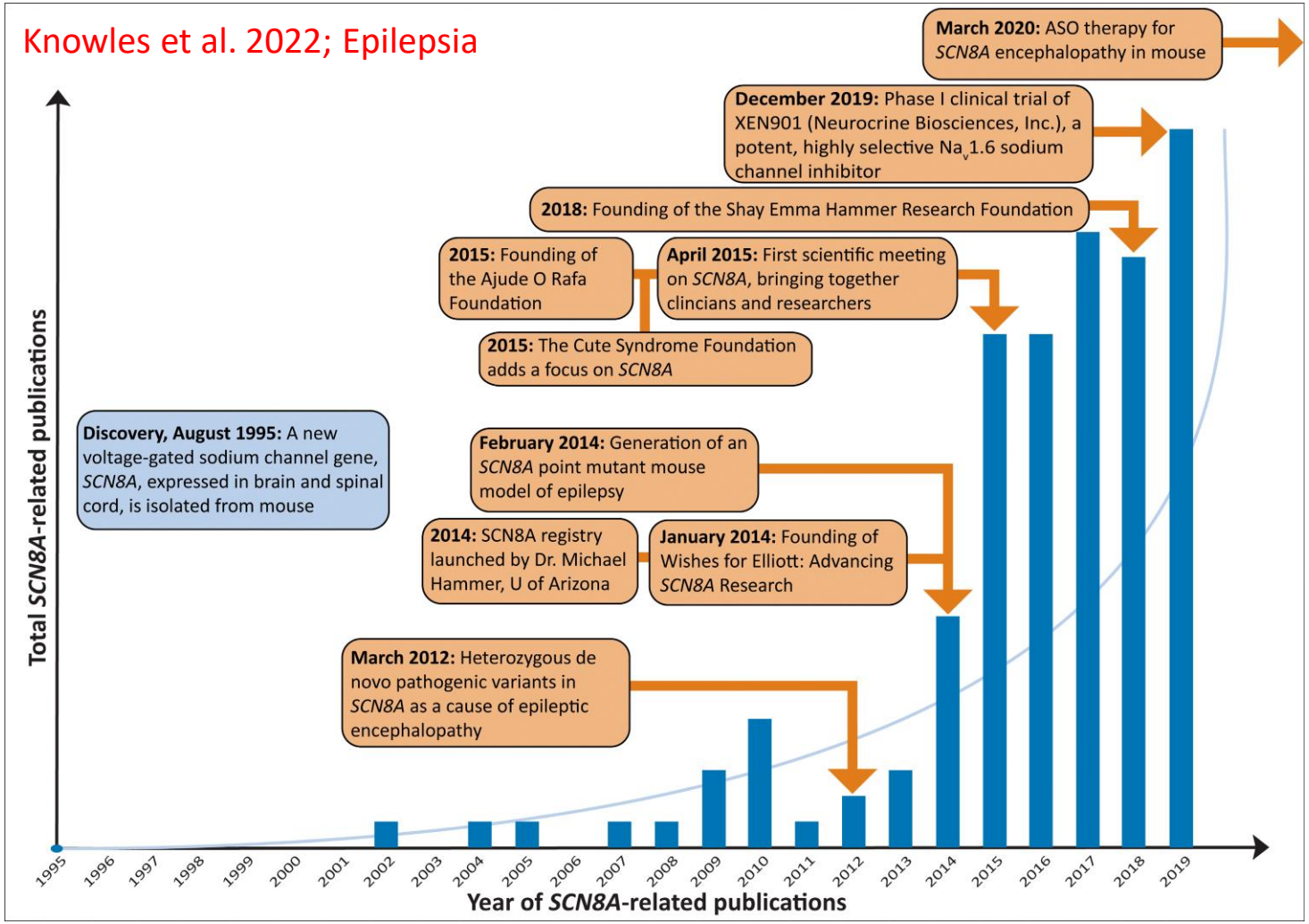
Facts & Challenges

Notwithstanding two decades of policy and legislation in Europe, aimed to foster research and development in rare conditions, only 5–6% of rare diseases have dedicated treatments.

With permission from the family

Theo is now in his 30's, completely cured following a long pre-surgical evaluation and epilepsy surgery

Facts & Challenges



Minimum 15 years from gene discovery to novel precision medicine approaches

Collection, validation, availability and sharing of DATA are PREREQUISITES

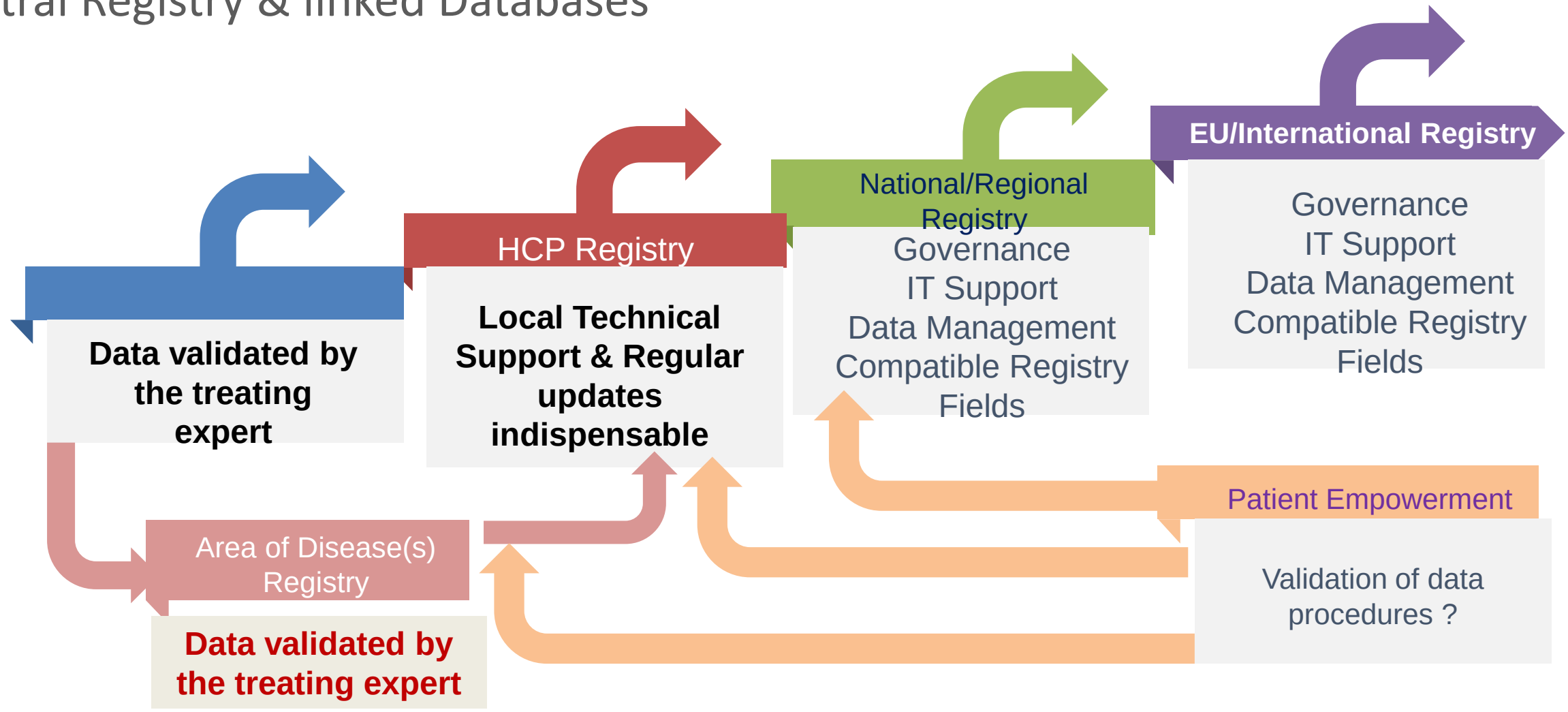
Take Home Messages - 2

2. Our biggest challenge relating to research and innovation is **the complexity of the diseases we are dealing with**.

It becomes urgent to adapt the regulations to the needs of the patients
as we cannot expect from the diseases to conform to the regulations.

The Data Sharing Challenge

A central Registry & linked Databases



The Data Sharing Challenge

A central Registry & linked Databases

Data validated by the treating expert

HCP Registry

Local Technical Support & Regular updates indispensable

National/Regional Registry

Governance
IT Support
Data Management
Compatible Registry Fields

EU/International Registry

Governance
IT Support
Data Management
Compatible Registry Fields

Area of Disease(s) Registry

Data validated by the treating expert

Patient Empowerment

Validation of data procedures ?

Sustainability - Updating

Take Home Messages - 3

3. Urgent to agree on regulations that **facilitate Data Sharing** at least between accredited medical teams, members of the ERNs.

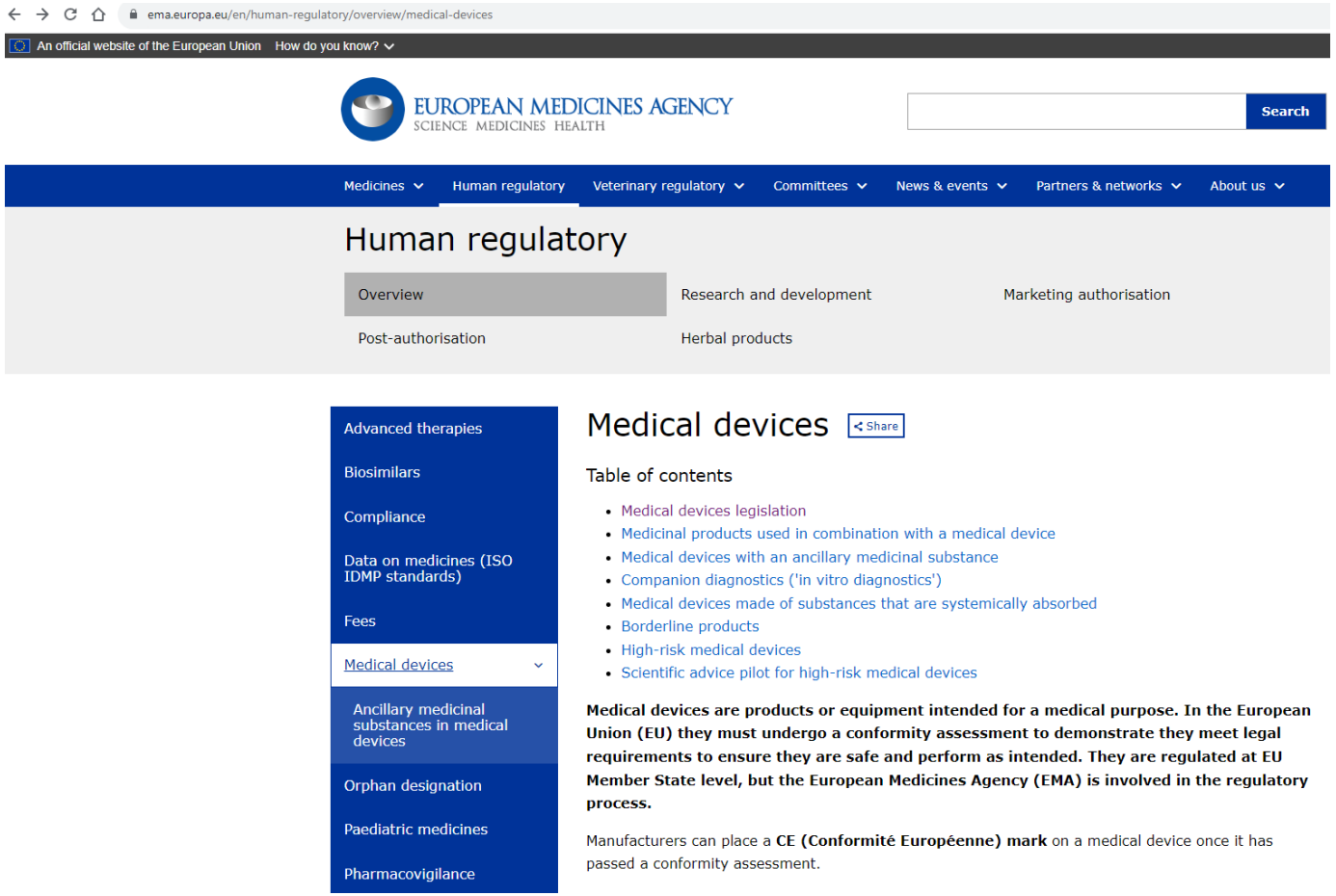
This remains highly complex even within the same country.

To achieve the maximum collection of data
let us start by facilitating the collection of the minimum

The Challenges

- Existing regulations make it **almost impossible to run non-sponsored clinical trials** in a rare disease.
  - ➢ The challenge to set up an EU/international ACADEMIC clinical trial for a rare entity, without the excessively costly involvement of a Contract Research Organizations.
- The competence of CROs in the domain of rare diseases.

Medical devices legislation



- The Regulation on Medical Devices (EU-2017/745) applies since 26 May 2021.
- The process is extremely time-consuming and expensive ALSO for devices which are NOT high-risk.
- For example, the approval of an App to help physicians in decision making is estimated to cost 0.5 million Euros.
- For-profit companies may be able to cover that, but many Apps are the outcome of non-for-profit clinical research and there is not funding for covering such huge expenses.

Take Home Messages - 4

4. The urgent need to **improve study designs** and **regulatory pathways** for the approval of medicines for ultrarare diseases, including repurposed medications.

Developing sustainable paradigms for making innovative medicines for ultrarare diseases broadly accessible and affordable to affected individuals and to societies.

The Challenges

- Discovery of new, secure, therapies (medical and surgical) require colossal funding. They can only be discovered (and really be targeted) after years of close collaboration between Pharma/Industry and the very small number of those that are knowledgeable in each of the rare diseases.

- **Collaboration**: to identify priorities; to increase our knowledge on the natural evolution of diseases; to understand the underlying mechanisms; and, based on the above, **define the most appropriate methodologies**.

Take Home Messages - 5

5. Rather than being first perceived as “conflicts of interest”,
transparent collaborations with industry
must be seen as the cornerstone of research & innovation,
at the interest of the patients.

EpiCARE Full & Affiliated members

The ERNs represent more than 1500 **“hand-picked” medical teams**,

accredited by their respective health authorities and the European Commission,

skilled in clinical research, respectful of already existing regulations that protect the rights of the patients, already collaborating with all stakeholders.

In 6 years they provided solid evidence of their ability to closely collaborate between them and with a large panel of patient advocates, progressively reestablishing a relationship based upon mutual trust.

Use the potential of the ERNs to urgently create,

in collaboration with them,

a legal framework that facilitates research and innovation