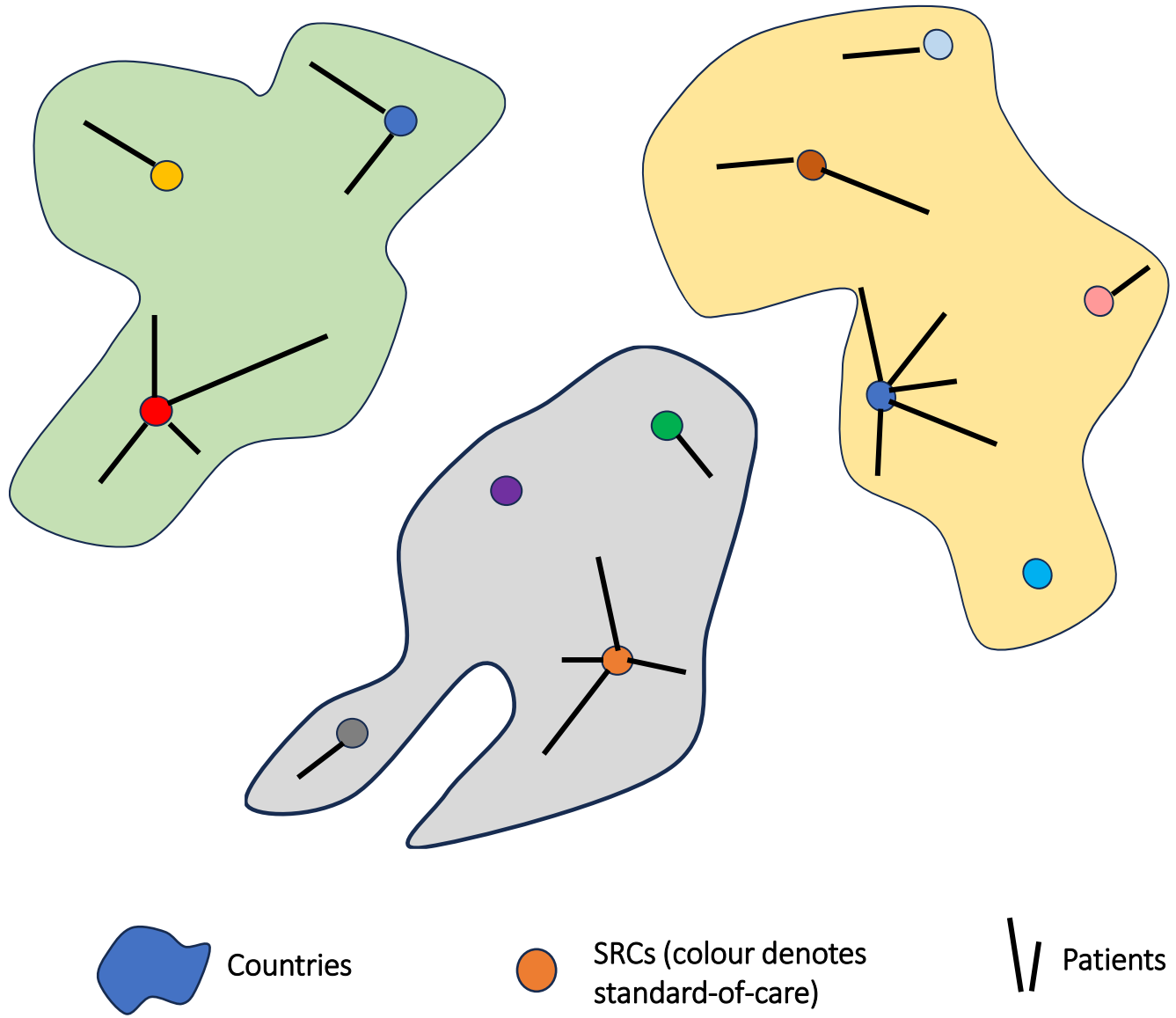
Ultra-rare sarcoma: Accessing patients

Hospital cohorts and mobilizing the patient community

Hugh Leonard
Chair of Trustees
The EHE Rare Cancer Charity

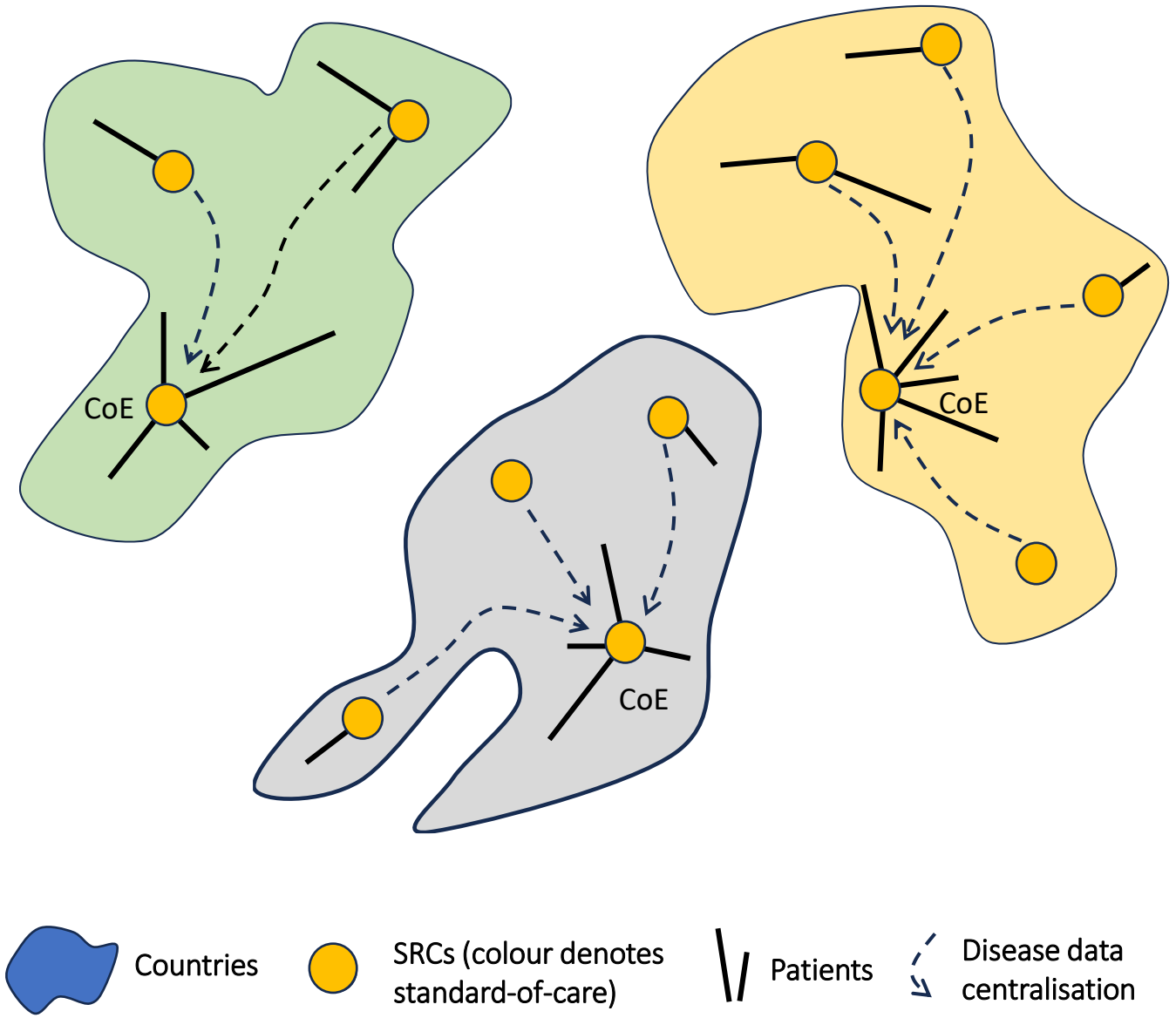
A typical picture for URSs?

- Some centres will have no patients
- Provisional centres may have 1 or2 patients
- Major population centres may have more
- Standard of care will be different in every centre;
- Recording of data will be different in every centre;
- Little appetite to start to standardise due to costs, tiny numbers, low priority
- Patients will be isolated and completely uncoordinated
- Collated data if achievable may be of little help in supporting drug approval processes
- Creates automatic rejection of retrospective data
- Massive inertia means the system continues as-is!



The dream?

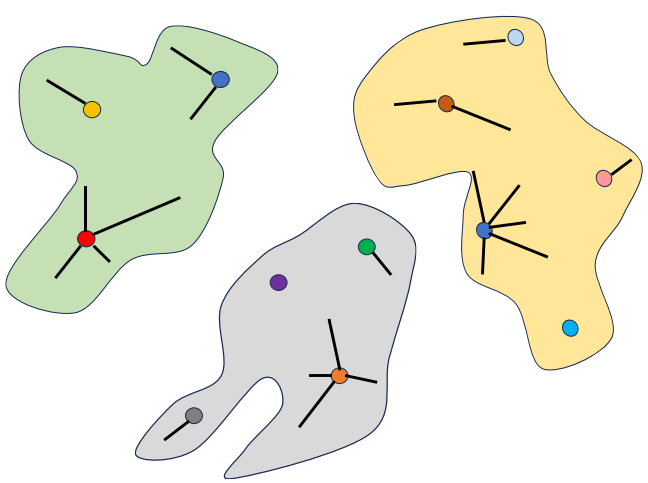
- Universal standard of care
- Standardised data collection and recording
- All data is collated at designated centres of excellence (CoEs) for the disease
- CoE maintains repository of all disease experience
- Maximises understanding and learning for clinicians
- CoEs carry obligation to feed-back learning to all SRCs ensuring better quality of care
- Biobanks may follow as care and processes become coordinated
- All data is now prospective and immediately useful for drug assessment and approval



Is transition realistic?

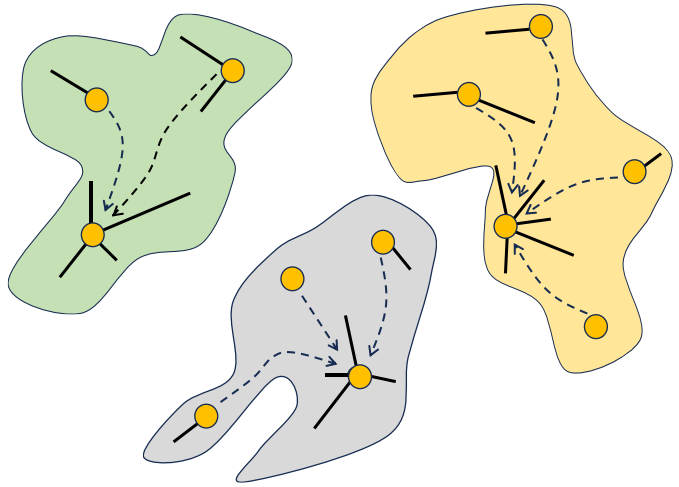
RARE CANCER CHARITY (UK)

YES!



So how do we get from this…

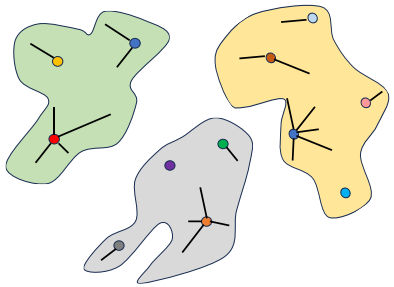
to this…



- By all stakeholders working together with common goals to deliver better treatment and better outcomes for the patient community
- Patients and their supporters are hugely motivated. Nobody is more vested in these outcomes than patients themselves

Mobilisation - The EHE Group story

……2012



- Isolated patients
- No coordination
- No collective voice
- No funding
- No attention
- No hope!

2013

- Started with 3 only
- More joined
- International interest
- Soon grew to 200+
- Great support
- Growing voice
- But no funds
- Better outlook?

2015

THE EHE FOUNDATION

RARE CANCER EHE CHARITY (UK)

RARE CANCER EHE FOUNDATION AUSTRALIA

- Over 500 members
- International engagement
- Fundraising started
- Funding of Rubin lab
- Engagement with clinicians
- Advisory Boards
- Growing voice
- Growing awareness

Today…..

CANADIAN EHE RESEARCH

EHE ITALIA
ASSOCIAZIONE NON SOLO LAURA

- **Over 2600 members**
- 85 countries
- **Over $10m raised**
- Multiple research funded
- Engaged with advocacy
- Delivering real world data
- Patient voices coordinated
- Demanding change and attention

The EHE research impact?

RARE CANCER EHE CHARITY (UK)

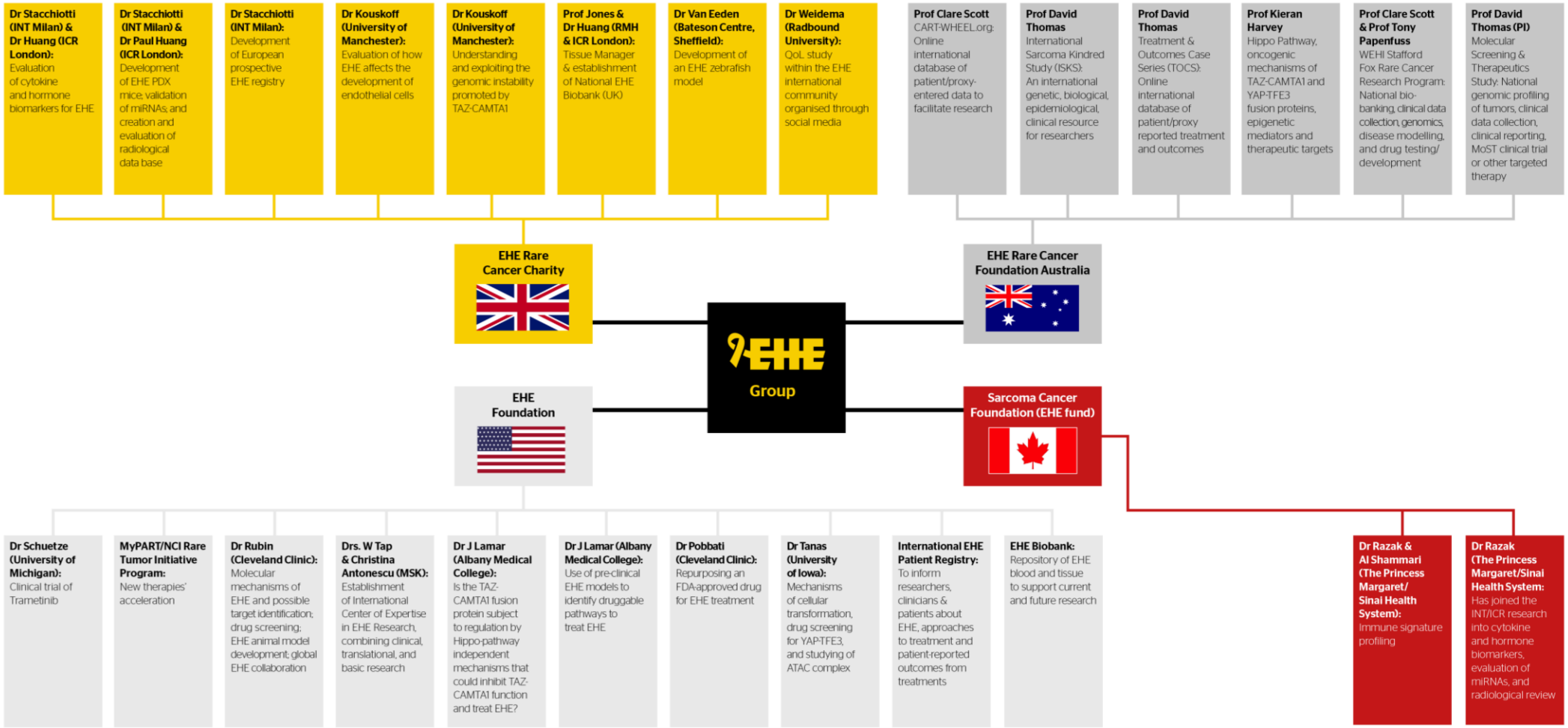
2015

USA

Dr Rubin (Cleveland Clinic): Working to identify the fusion protein associated with EHE; seeking to establish a mouse model

The EHE research picture today – 26 projects

RARE CANCER EHE CHARITY (UK)



Patient contributions to disease research

RARE CANCER EHE CHARITY (UK)

- The EHE Group has already helped deliver the following, with funding and/or patient engagement:
  - Identification of the **EHE fusion proteins** (TAZ-CAMTA1 and YAP-TFE3),
  - Importance and mechanism of **TEAD binding**;
  - The possible role of **DNA damage** and **HR corruption**;
  - A robust **GEM model** of EHE;
  - Pluripotent **stem cell model** of EHE;
  - Other models including **human cell lines** and **zebrafish**;
  - Identification of likely **biomarkers**;
  - Major **observational study** of EHE (50 participants)
  - World first multi-centre **prospective registry** (22 centres)
  - **EHE biobanks** in the UK and USA;
  - **Global EHE patient registry** delivering real world data
  - Patient engagement with **clinical trials** (Mekinist and TEAD-inhibitors)

- In one word - **MOMENTUM**

The patient community is a powerful partner

RARE CANCER EHE CHARITY (UK)

- The patient community is globally coordinated, mobilised and hugely motivated to deliver successful disease research and progress - a powerful and valuable partner;
- We will continue to promote and encourage patient engagement with all EHE registries, studies and other key data gathering processes;
- But the patient community does reasonably expect:
  - To be fully involved and for their voice to be genuinely heard. They will not be ignored!
  - All parties to work together to ensure that established procedures based on common disease practice are not an impediment to progress, no matter how comfortable we are using them;
  - All appropriate steps to be taken to minimise or remove rarity and limited commercial potential as an impediment to progress;
  - The very significant and unmet need of ultra-rare disease patients to be genuinely recognised and all elements to be handled with appropriate **urgency**;
  - All involved parties to work openly and diligently to find workable solutions and deliver the treatments so badly needed.

Working together to optimise patient contributions

RARE CANCER CHARITY (UK)

- So patients are ready to engage, but we need to work hard to optimise their contributions
- We must maximise what we can get from each patient, in quantity, quality and timing
- We will need to use and/or develop the appropriate infrastructure to allow us to:
  - reach and engage patients;
  - capture and collate data;
  - involve researchers and academia in analysis and evaluation of diseases and drugs; and
  - ultimately deliver to regulators what they require to support drug approval applications
- Critically, we need to agree in advance with stake holders, **particularly with regulators**, what data needs to be collected to support drug assessment and approval
- We also want and need to agree this with all key regulators together so that when we collect data, we know it will support all the main jurisdictions
  - Going through this process with each regulator individually is just not sensible!

Our collective responsibility!

- Do we have the collective ambition to deliver a new and appropriate drug approval pathway for ultra-rare diseases that is so important to the patient communities?

- Can we establish a real time, working group structure, involving key stakeholders, to deliver this important objective?