RARE DISEASE PATIENTS' NEEDS AND GOALS

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Where are we coming from? Progresses in the last ten years

European
Reference
Networks

2008

2009

2011

2017

EUCERD
2010-2013

EC Expert
Group on RD
2013-2016

EUCERD Joint Action
(2012-2015)

RD-ACTION
(2015-2018)

But where are we really at?

24 National Plans or Strategies for Rare Diseases (only 4 in 2008)

Uneven or scarce implementation

Inequalities across EU

Thanks to ERNs, rare diseases are at the forefront of health policy

ERNs are still disconnected from **national healthcare systems** and **patient pathways**

Social & healthcare systems largely separated

New scientific developments, new therapies & new diagnostic landscape

New **challenges in pricing & access** to therapies

Existing **pricing models not sustainable**

What do we need to do?

Major **achievements** compared to 20 years ago but still significant challenges

Individual countries cannot face **alone** issues related to Rare Diseases

We need a **new policy framework** with strong EU support and collaboration between Member States

We need to identify **key policy priorities** to address outstanding issues in an integrated manner

- We **asked the National Alliances**
- We want to ask the **overall RD community → you today**

How we worked

Objective: Define key priorities for the next decade in order to create favourable environment for a EU-national policy framework.

→ Brainstorming

→ Written consultations

→ Debates

→ Final survey

- Starting from the policy areas of the *2008 Commission Communication* and the *2009 Council Recommendation* and *EUROPLAN national conferences*
- Building on EURORDIS position papers developed with the members' contribution

How did we assess priorities

→ Initial kick-off and discussion with Council of National Alliances across the whole spectrum of RD-related issues

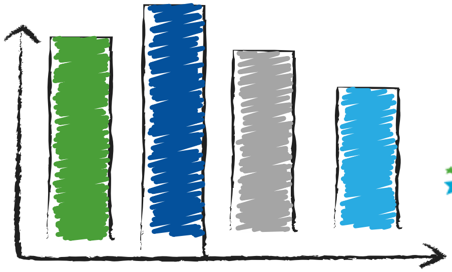
→ Creation of a **Working Group on future RD Policy Priorities**

Alba Ancochea – FEDER – Rare Diseases Spain
Claudio Ales & Simona Bellagambi – UNIAMO – Rare Diseases Italy
Eleni Antoniou – CARD - Rare Diseases Cyprus
Eleni Paximadi – PESPA – Rare Diseases Greece
Geske Wehr – ACHSE – Rare Diseases Germany
Ivana Badnjarevic – NORBS - Rare Diseases Serbia
Lene Jensen & Birthe Holm – RDD - Rare Diseases Denmark
Nick Meade - UK Genetic Alliance/ Rare Disease UK

→ Presentation and discussion on selected priorities with Council of National Alliances to identify gaps and key "asks"

→ **Survey on KEY policy priorities** with all National Alliances

→ **How to prioritize between necessities?**



Future priorities for rare diseases in an integrated ecosystem

CROSS-CUTTING

Patient engagement & empowerment

Education of all actors in the field

EU – NATIONAL LEVELS

EU guidance

National Plans and Strategies

BUILDING BLOCKS

(codification, data sharing and collection standards, interoperability & IT tools)

Future priorities for rare diseases in an integrate ecosystem

Research

Access to Treatments

Diagnosis and Healthcare

Social Care

Research for rare diseases

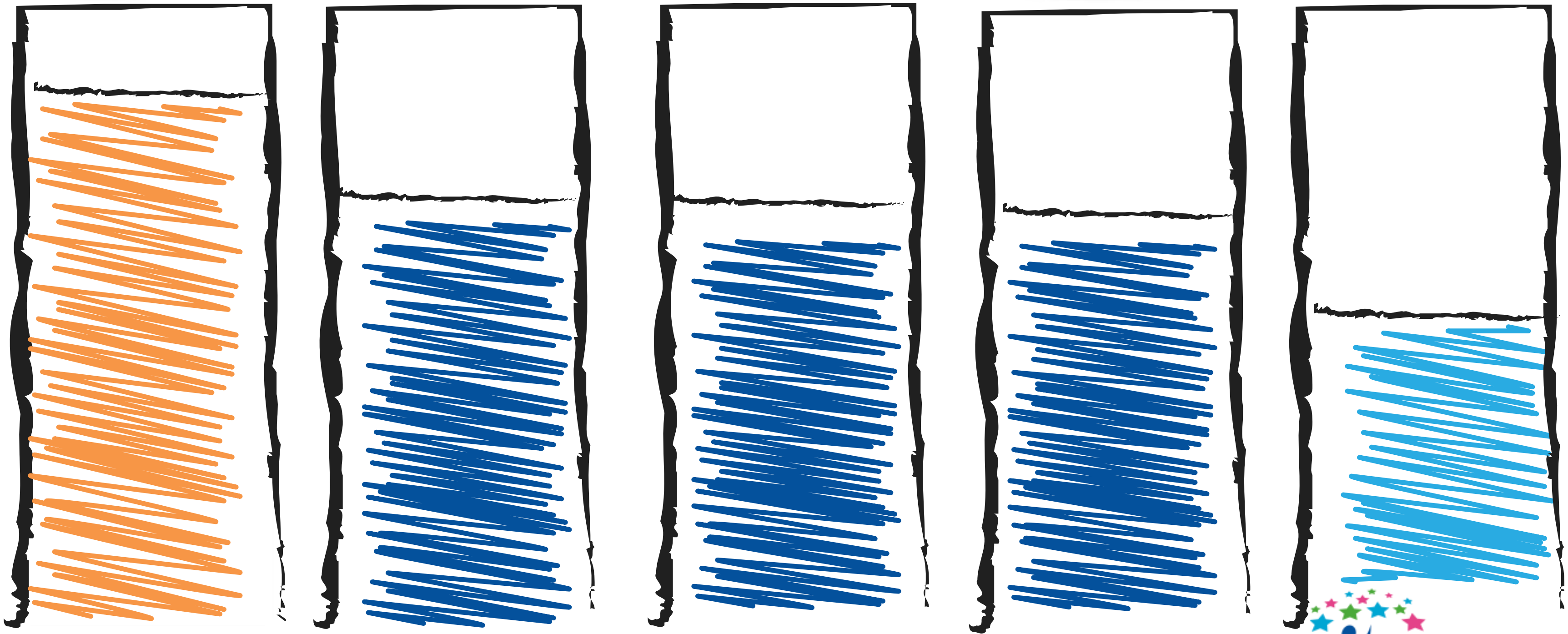
Systematic collection of findings from diagnostic & care

Developing Clinical Research Networks from ERNs

Horizon scanning & gap analysis of unmet needs

Social research and research on QoL

Coordination of research infrastructures



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Development and Access to Treatments for RD

Patient Reported Outcomes & Real World Evidence

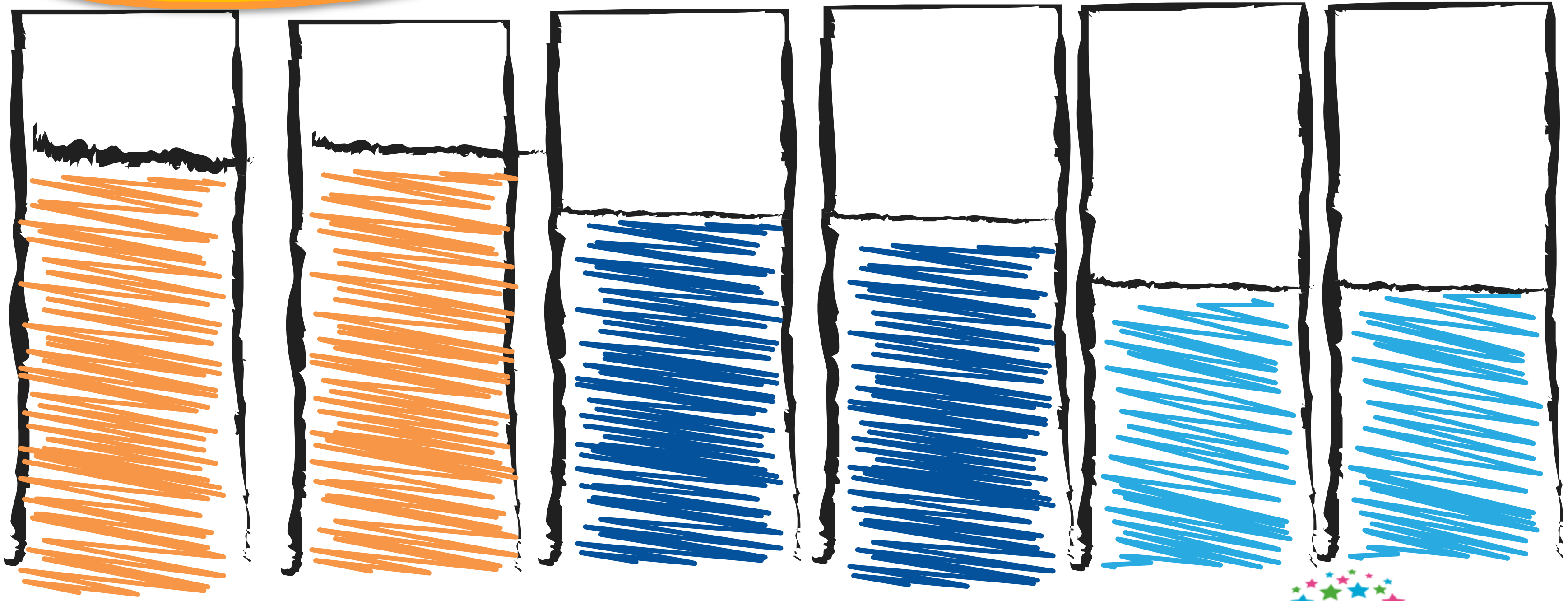
Joint clinical reports for **Health Technology Assessment**

Adaptive regulatory pathways

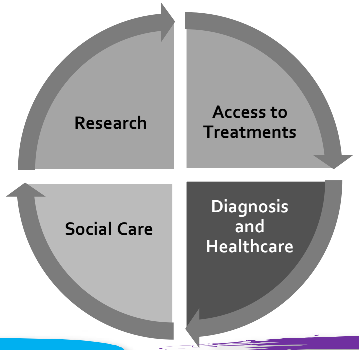
Innovative clinical trial designs

EU Table of Negotiation on pricing of treatments

EU Fund for post marketing evidence generation



Improving Diagnosis & Care for RD


Research

Access to Treatments

Social Care

Diagnosis and Healthcare

Primary care referral to specialists for right diagnosis and/or care

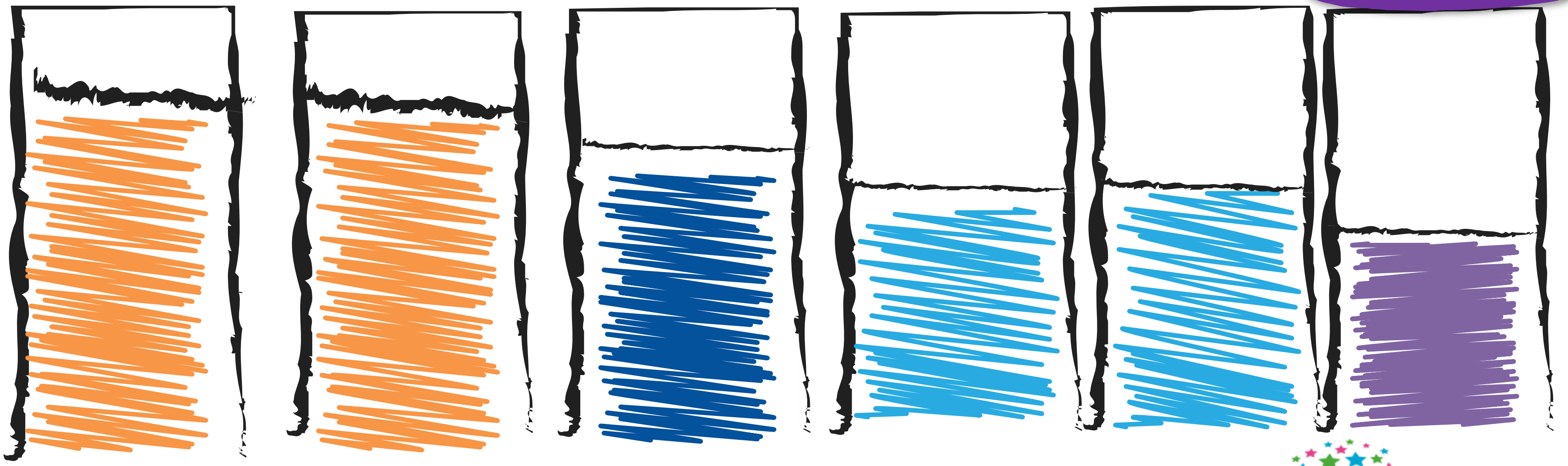
Deploying **ERNs** & **anchoring** into national healthcare systems

Reinforcing collaboration o **undiagnosed diseases**

Cross-border access to diagnostic tests

Wider use of new-born screening

Strengthened **EU work** to implement **cross-border patients rights**



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Social Care for RD

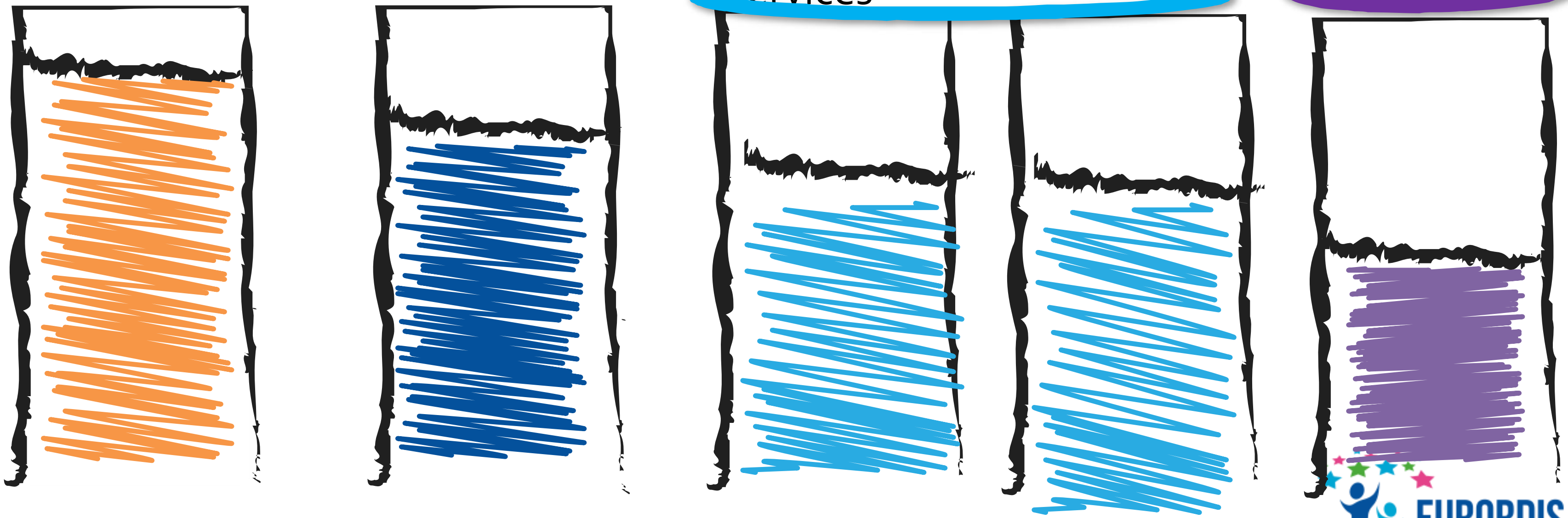
Mechanisms to **support integrated care** for RDs

Integration of RDs into **mainstream** services and policies

Provisions of social care for RDs in specialised health services

Work-life balance

Specialised social services for RDs



Future priorities for rare diseases in an integrate ecosystem

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Access to Treatments

Diagnosis and Healthcare

Social Care

BUILDING BLOCKS

(codification, data sharing and collection standards, interoperability & IT tools)

Consolidating Building Blocks

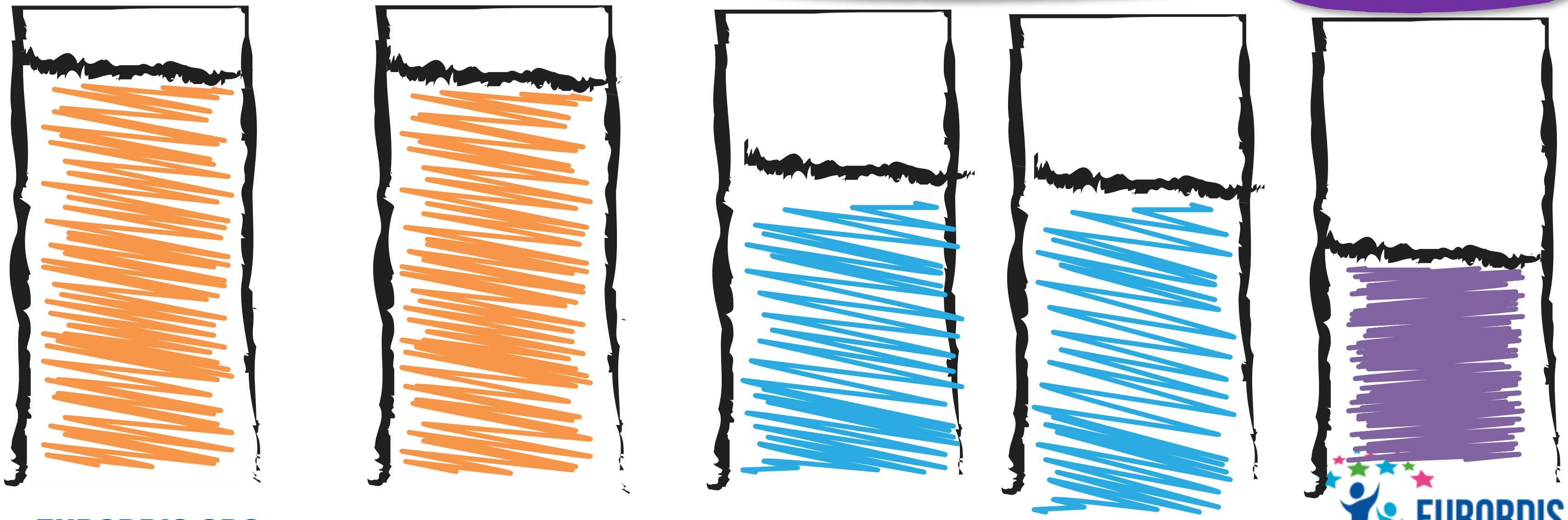
Traceability of rare disease patient data across national healthcare systems

Wider systematic use of **Orpha Codes**

Systematic collection of quality data at key settings

Interoperability of **registries, databases, EHR**

Data quality & reliability of information collected by **mHealth** apps



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Cross-cutting & overarching issues

Active engagement and active role of patients
@ all key decision-making points

Adoption, Implementation & monitoring of **National Plans or Strategies**

Education & training of ALL actors

We asked the National Alliances – now we ask you!

→ We want to ask the **overall RD community → you today.**

→ **Choose two out of the "number ones" from the four themes:**

- **Systematic collection** of findings from diagnostic & care.
- **Patient Reported Outcomes & Real World Evidence**
- Joint clinical reports for **Health Technology Assessment**
- **Primary care referral** to specialists for right diagnosis and/or care
- Deploying **ERNs** & **anchoring** into national healthcare systems
- Mechanisms to support integrated care for RDs

The way forward for people with rare diseases

What can people with RD actively do?

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THANK YOU

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