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EUROPEAN PAEDIATRIC TRANSLATIONAL RESEARCH INFRASTRUCTURE

THE HIGHEST CHALLENGE OF RARE DISEASES IN CHILDREN

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Rare Diseases (RD)

- Group of **7,000 conditions**
- Affect approximately **60 million people in the US and Europe**
- The majority of RD have a **genetic origin**
- Many rare diseases are **chronic and life threatening**



Rare Diseases in Children

- Three-quarters of RD also affect children (genetic origin)
- Many RD begin in childhood and are chronic conditions
- Multidisciplinary approach is the gold-standard in the pediatric setting



Pediatric Research Field

European Union Paediatric Regulation since January 2007:

- Improve the **health of children** in Europe
- Facilitate the **development and availability of medicines** for children aged 0 to 17 years
- Ensure that medicines for use in children are of **high quality, ethically researched** and **authorised appropriately**



Rare Diseases and Pediatric Research Field

- It is estimated that 94% of RD continue to **lack an approved and available therapy**
- Few (22%) of these conditions have been studied in drug trials
- **Children may be particularly vulnerable** to the challenges of conducting clinical trials for RD
- There is **no simple way to insure that children benefit** from pharmaceutical progress
- Development of a **new drug** has **high costs**



Rare Diseases Trials in the Pediatric Field

CHALLENGES:

- Disease rarity = small populations
- Geographic dispersion of patient populations
- Difficulty recruiting participants
- Clinical heterogeneity
- Limited knowledge of natural disease history
- Inaccurate diagnosis
- Lack of validated clinical parameters
- Lack of harmonized disease registries
- Limited disease experience in the medical community



The High Needs of RD

- Evidence from **basic research and preclinical research**
- **Evidence-based clinical practice:**
 - harmonized databases and registries
 - measurable patient-centred outcomes
 - appropriate study designs adapted to
 - small study populations
- **Research Infrastructures**
- **Reference Expertise Networks (ERN BOND)**
- Improve **public awareness** about RD



The High Needs of RD

- **Harmonized regulatory procedures** at EU level
- Regulatory and financial incentives **promoting drug development**
- New innovative drugs and therapeutic concepts for **further breakthroughs**
- Increase rigorous **trial activity** adapted to the intrinsic complexity of RD
- Increase **multi-stakeholder engagement** (patients, scientific community, industry, and policymakers)

The Highest Challenge of RD Field in Children

Guarantee the highest positive impact on the quality of life of children with RD as early as possible.



CONCLUSIONS

- **Many RD begin in childhood** and are chronic conditions.
- There is a **need for evidence** from basic to clinical research in RD.
- There are **many challenges** in the RD research, especially in the pediatric field.
- National and International **Reference Networks** are very helpful.
- Strong **partnership between stakeholders** are key for success.





European
Reference
Network

for rare or low prevalence
complex diseases



Network

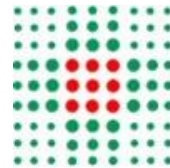
Bone Disorders (ERN BOND)



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THANK YOU!

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