HORIZON EUROPE (2021 – 2027)

Investing to shape our future

The current state of research into rare diseases in the EU

Conference on rare diseases & ERNs
11 October 2023 - Bilbao

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European Commission

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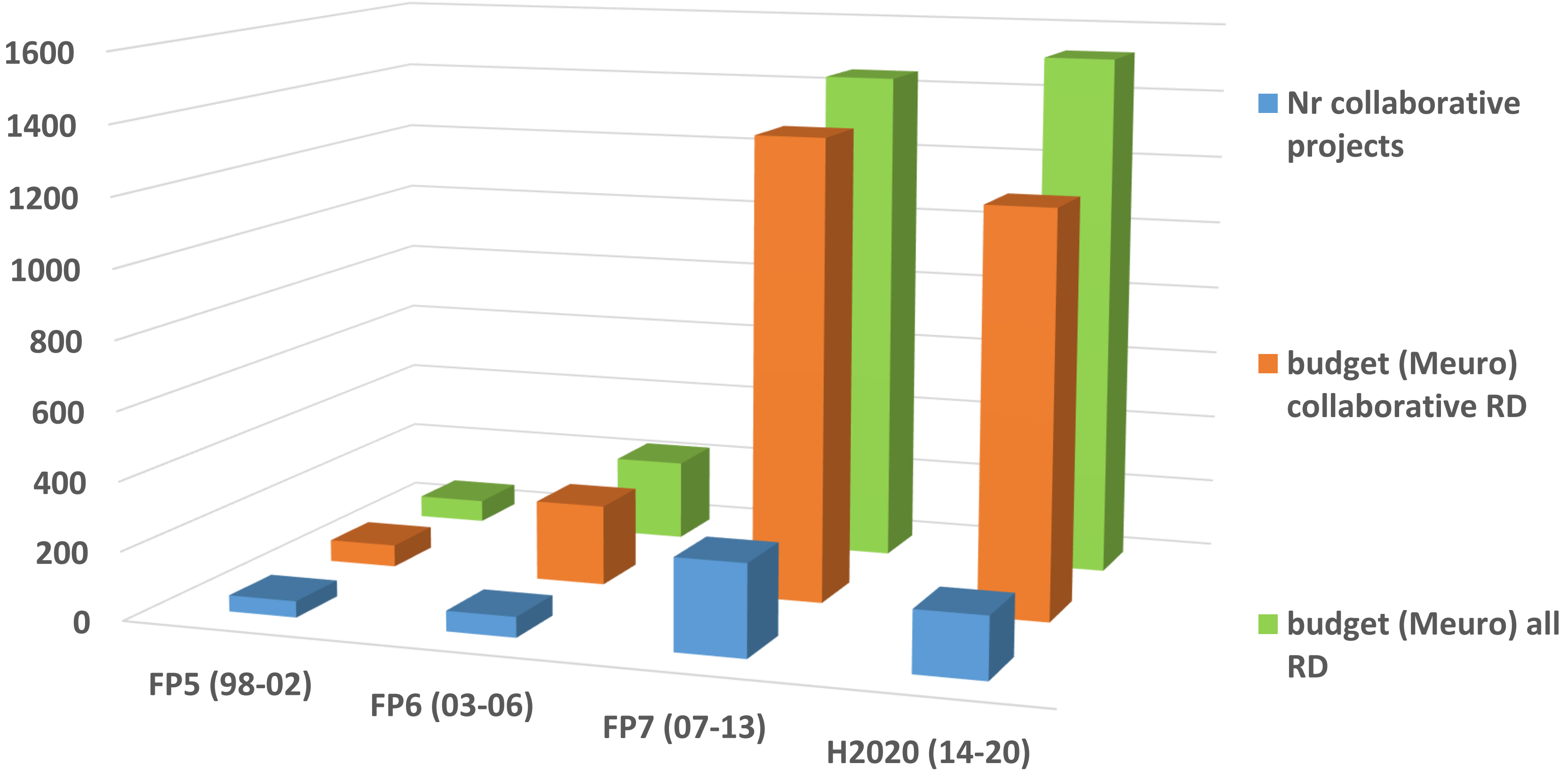
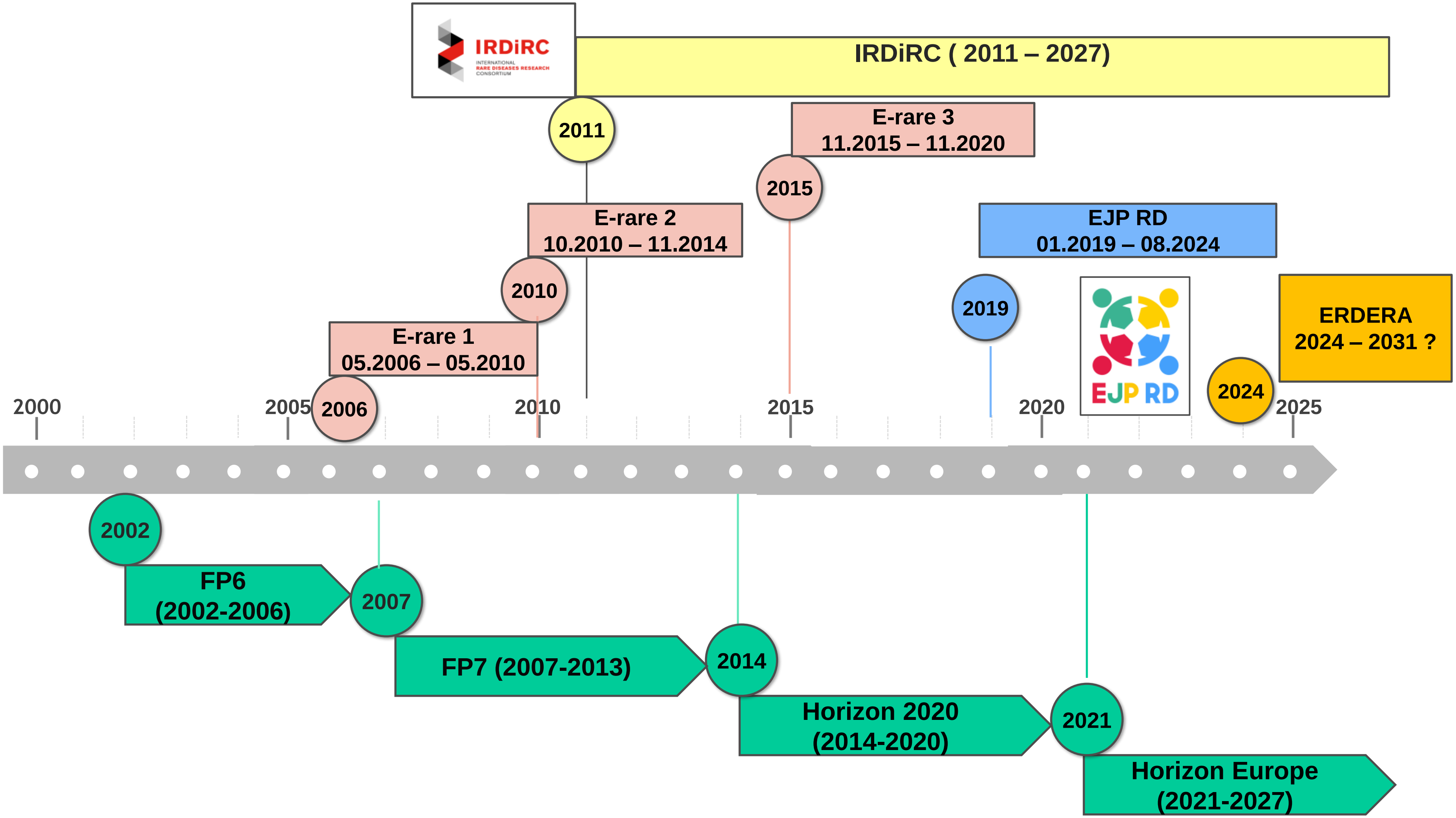
EU Research & Innovation on Rare Diseases
a long-standing commitment from funding programs



EU research & innovation on Rare Diseases
Coordination of national & international research funding



IRDiRC INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

IRDiRC (2011 – 2027)

2011

E-rare 3
11.2015 – 11.2020

2015

E-rare 2
10.2010 – 11.2014

2010

EJP RD
01.2019 – 08.2024

2019

EJP RD

ERDERA
2024 – 2031 ?

2024

E-rare 1
05.2006 – 05.2010

2006

2002

FP6
(2002-2006)

2007

FP7 (2007-2013)

2014

Horizon 2020
(2014-2020)

2021

Horizon Europe
(2021-2027)

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The International Rare Diseases Research Consortium (IRDiRC) a key player to put RD research on the agenda at global level

IRDiRC – Vision and Goals by 2027

Vision

Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.

Goals

- All patients coming to medical attention with a suspected rare disease will be **diagnosed** within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline
- 1000 new **therapies** for rare diseases will be approved, the majority of which will focus on diseases without approved options
- **Methodologies will be developed to assess the impact** of diagnoses and therapies on rare disease patient

European Joint Programme on Rare Diseases (2019-2024) Main achievements

Accelerating of research translation & clinical studies

Innovation Management Toolbox – 450 resources, 15 use cases

Mentoring programme: 28 projects, 42 experts DB, over 500 hours of mentoring

CT toolbox kit

5 Methodological innovation & validation projects

Coordination & transversal activities

Qualified coordination team & support

Agile governance & strategy

Sustainability planning from 1st day – business plan developed

Extensive ethics & regulatory support

Perfomant communication & dissemination

Central Helpdesk – 450 experts

RD research funding

4 JTCs – 83.5 M€ - 64 projects – 432 research teams

45 Networking events – 1.24 M€ - 43 topics covered – 33 countries including 15 underrepresented

3 RD Research public-private challenges (1.5 M€)

78% funded projects involve patient organisations

Capacity building & empowerment

RD data management & quality trainings – 892 trainees

Patient trainings (therapy dev., RD research, leadership, young patients) – 440 trainees

31 ERN workshop financed

89 ERN fellowships attributed

2 Online education MOOCs (diagnosis & translational research) – 8704 trainees from 148 countries

EJP RD

www.ejprarediseases.org

Access to data, tools & services for research

Virtual Platform set up: https://vp.ejprarediseases.org

22 resources connected (11 registries, 3 catalogues, 2 gen-phen deposition infras, 5 knowledge bases, 1 project)

4 levels of onboarding

Over 100 biological pathways created

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* Slide: courtesy Daria Julkowska, INSERM, EJP RD coordinator

ERICA: The European Rare Diseases Research coordination and support action to support clinical research efforts of ERNs

www.erica-rd.eu (2021 – 2025)

ERICA
European Rare Disease Research
Coordination and Support Action

Objectives: to increase the clinical research and innovation potential of European Reference Networks (ERNs), through:

- facilitating inter-ERN collaboration,
- increasing the visibility and impact of ERNs (outreach and collaboration with multiple stakeholders)

Work Packages (WPs)

- wp1 Consortium management and coordination
- wp2 Data Collection, Integration and Sharing
- wp3 Patient-Centred Research
- wp4 Clinical Trial Support
- wp5 Translation and Innovation

- wp4 Clinical Trial Support
- wp5 Translation and Innovation

- wp6 Integration, Outreach & Dissemination
- wp7 Ethics Requirements

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ERNs & their registries: a huge potential !

ERN	ERN registry project
Endo-ERN	EuRRECa
ERKNet	ERK-Reg
ERN-LUNG	REGISTRY WAREHOUSE
MetabERN	U-IMD
ERN PaedCan	PARTNER
ERN GUARD-HEART	GUARD-Heart Registry
ERN ReCONNET	TogethERN ReCONNET
ERN ITHACA	ILIAD
ERN BOND	EuRR-Bone
ERN CRANIO	ERN CRANIO registry
ERN EURACAN	STARTER
ERN RND	ERN-RND Registry

ERN	ERN registry project
ERNICA	ERNICA registry
ERN EYE	REDgistry
ERN EpiCARE	EPICARE-GRANT
VASCERN	VASCERN Registries
ERN-Skin	ERN-Skin REGISTRY
ERN EURO-NMD	EURO-NMD Registry
ERN GENTURIS	GENTURIS registry
ERN RARE-LIVER	R-LIVER
ERN RITA	MERITA
ERN TRANSPLANT-CHILD	PETER
ERN EuroBloodNet	ENROL
ERN eUROGEN	ERN-eUROGEN registry

Joint Research Centre
JRC

European Platform on Rare Disease Registration (EU RD Platform)

Searchable, findable rare disease registry data

https://eu-rd-platform.jrc.ec.europa.eu/erdri_en

Solve-RD: Solving the unsolved rare diseases

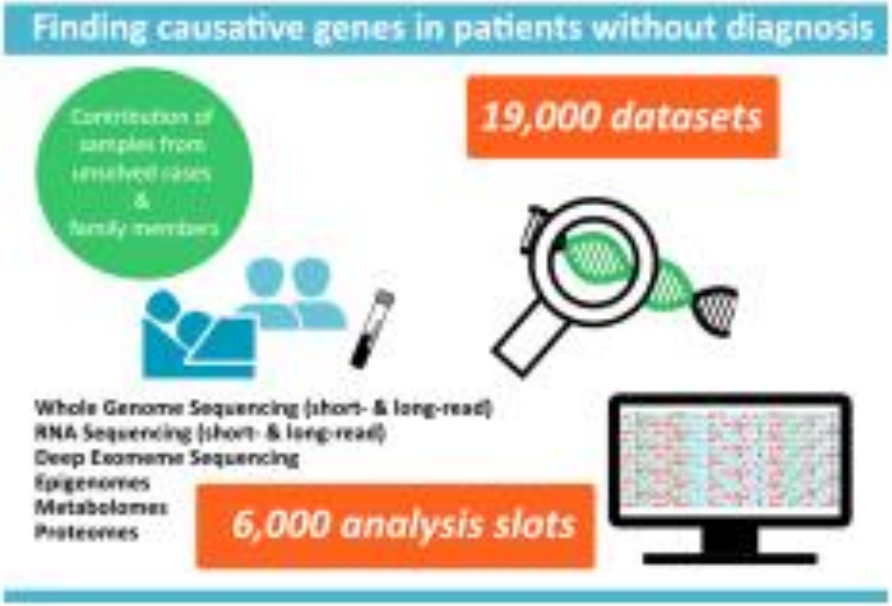
www.solve-rd.eu (2018 – 2024)

Solve RD

Re-analysis of unsolved exomes and genomes

Novel omics approaches

< 50 groups in 2018; > 200 groups in 2022



Solve-RD organises 3 “Solvathons” - data analysis and interpretation workshops on different topics (Nov. 2023, Feb. & April 2024)

* Slide: courtesy Dr Holm Graeßner, Uni Tuebingen, coordinator Solve-RD

Innovative Medicines Initiative (IMI) – Horizon 2020
Innovative Health Initiative (IHI) – Horizon Europe

IMI:

- **C4C ‘conect4children’ - Collaborative network for European clinical trials for children’ (2018 – 2024)** **€182 mio**

- **Screen4Care ‘Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies’ (2021 – 2026)** **€25.5 million**

IHI ongoing call:

- **Topic 4: Establishing novel approaches to improve clinical trials for rare and ultra-rare diseases**
- (Opened on 27 July 2023, 2 stages: Deadline: 8 Nov 2023 & 23 April 2024)

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Horizon Europe – Health Cluster calls (2021-2022)
Development of new effective therapies for Rare Diseases

EU RAS	EUropean network for neurodevelopmental RASopathies
GEREMY	Gene Therapy for treatment of rare inherited Arrhythmogenic Cardiomyopathy
MAGIC	Next-generation models and genetic therapies for rare neuromuscular diseases
NANEMIAR	Nanomedicine Approach to Normalize Erythrocyte Maturation in Congenital Anemia by messenger RNA
RESTORE VISION	Novel advanced and repurposed therapeutics for vision restoration in a group of severe rare ocular surface diseases: from validation to first clinical investigations
SIMPATHIC	Accelerating drug repurposing for rare neurological, neurometabolic and neuromuscular disorders by exploiting SIMilarities in clinical and molecular PATHology
TheRaCil	Therapies for Renal Ciliopathies

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Horizon Europe – Health Cluster calls (2021-2024)

HORIZON-HLTH-2022-TOOL-11-02: New methods for the effective use of **real-world data and/or synthetic data** in regulatory decision-making and/or in health technology assessment

→ **Real4REg:** Development, optimisation and implementation of artificial intelligence methods for RWD analyses in regulatory decision-making and health technology assessment along the product lifecycle Jan 23 – Dec 26

HORIZON-HLTH-2023-IND-06-04: **Modelling and simulation to address regulatory needs in the development of orphan and paediatric medicines**
Deadline**: 13 April 2023** Instrument: RIA

→ Under grant preparation –
new projects expected to start Q1 2024

HORIZON-HLTH-2023-IND-06-05: **Mapping the hurdles for the clinical applications of Advanced Therapy Medicinal Products (ATMPs)**
Closes: **13 April 2023** Instrument: CSA

→ Under grant preparation –
new projects expected to start Q1 2024

ERDERA (European Rare Diseases Research Alliance) proposed co-funded European Partnership on Rare Diseases

HORIZON-HLTH-2023-DISEASE-07-01
European Partnership on Rare Diseases
Closes: **19 September 2023**
Instrument: HORIZON-COFUND

Indicative budget: 50M€ (2023)
Maximum budget: 150M€ (with 2nd instalment in HE Work Programme)

But don't forget the other Partnerships in cluster Health:

- **ERA4Health** – started in 2022
- **Transforming Health & Care Systems (THCS)** – launched early 2023
- **Personalised Medicine** – will start end 2023

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https://europa.eu/!wXBg9v

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ERDERA (European Rare Diseases Research Alliance)

Novelties compared to EJP RD

- Grow & consolidate a multi-stakeholder R&I ecosystem
- > 50% budget to Community-building actions and integrative activities;
  - Strengthening the European Research Area on rare diseases R&i
  - The Clinical Research Network for clinical trial preparedness
  - Diagnostic pipeline for solving
  - Implement multicountry clinical trials
  - Boost public-private collaboration
  - Accelerate ATMPs development

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COLLABORATION:
A KEY TO UNLOCK
THE CHALLENGES OF
RARE DISEASES RESEARCH

FEBRUARY 2021

Rare diseases

A staggering 6000 to 8000 life-threatening, or chronically debilitating, rare diseases that each one affect less than 5 in 10,000 persons. It is estimated that altogether rare diseases affect more than 30 million people in the European Union.

Many rare diseases manifest themselves in childhood, resulting in a shortened lifespan and leading to a dependency on care throughout the patients' lives, causing significant suffering to the patients and their families. Patients affected by rare diseases often spend years enduring a "diagnostic odyssey" before receiving the correct diagnosis, if ever.

Most rare diseases lack effective treatments representing an enormous unmet medical need. Rare disease patients often need highly specialised health care and social services resulting in high costs for their families and for society.

Each rare disease affects a small number of people, each with its specificities, leading to scarcity and fragmentation of knowledge and expertise. This is why rare diseases are recognised as a field where European and international collaboration is indispensable to improve diagnosis and find treatments.

EU funded research on rare diseases

The EU facilitates the formation of multidisciplinary consortia with participants from universities, research organisations, healthcare providers, SMEs, industry and patient organisations from across Europe and beyond.

Over the past 14 years, the EU has supported this field extensively through its Framework Programmes for Research and Innovation with more than €2.4 billion attributed to over 800 research and innovation projects. Research on rare diseases is supported by FP7 (2007-2013) and continues to be supported in Horizon 2020 (2014-2020) in various ways with the majority of the funds directed towards collaborative research projects (consortia) under the Health theme and the Health Societal Challenge respectively. Furthermore, the European Research Council (ERC), the Marie Skłodowska-Curie Actions and the European research infrastructures programmes

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Thank you!

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http://ec.europa.eu/horizon-europe

https://ec.europa.eu/info/research-and-innovation/research-area/health-research-and-innovation/rare-diseases_en

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