European research on rare diseases (RD)

a long-standing commitment to make Europe a leader at global level

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DG Research & Innovation, European Commission

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Framing the ecosystem

with coordinating initiatives

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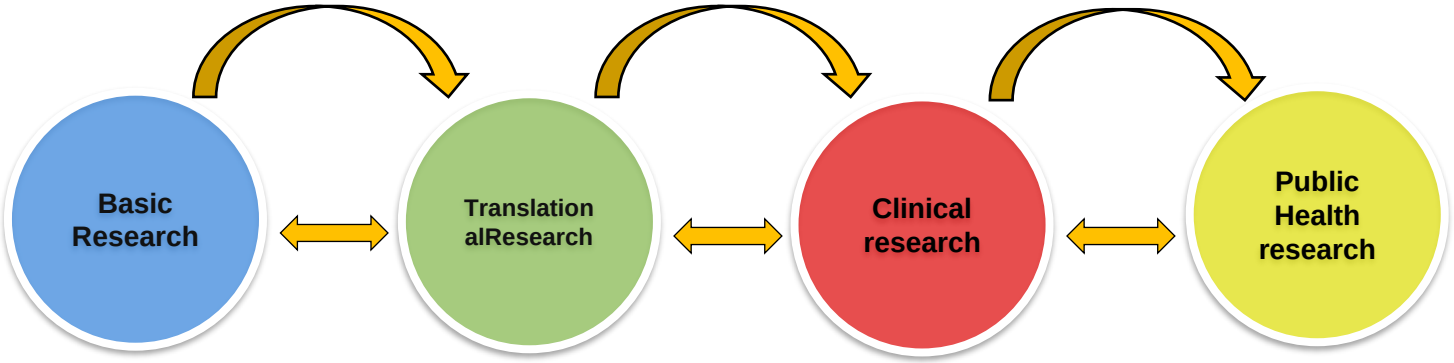
EU collaboration on rare diseases
Targeting & integrating different stages of the research & innovation path



In 2024, we celebrated the 40th anniversary of the EU's first funding programme dedicated to science, research and innovation. It all started in 1984 with the launch of the Framework Programme for Research and Technological Development…

Celebrating 40 years of EU Research and Innovation - European Commission

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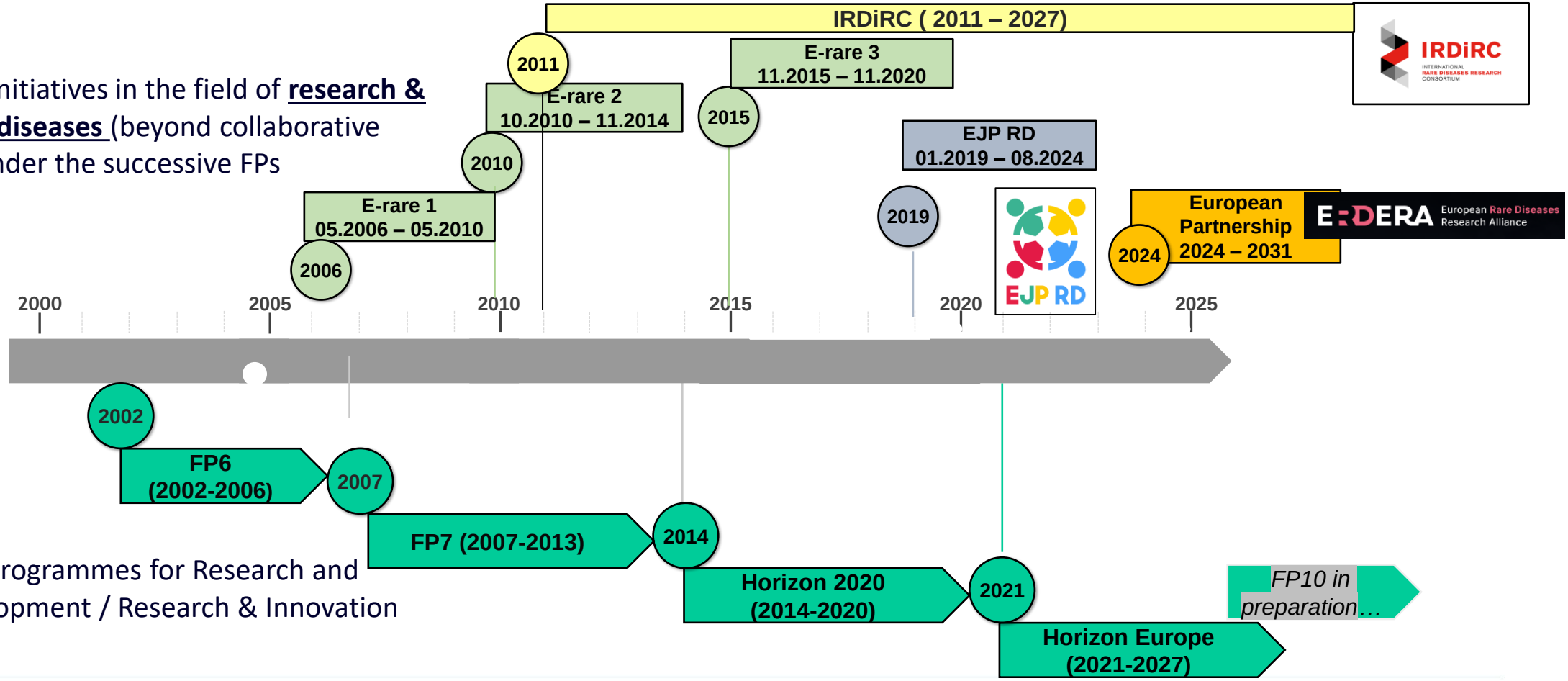
EU research & innovation activities on rare diseases: Coordination of national & international research funding

Main coordinating initiatives in the field of **research & innovation on rare diseases** (beyond collaborative research) funded under the successive FPs



FP: EU Framework Programmes for Research and Technological Development / Research & Innovation

EU investment on RD research : complementary funding models targeting different research needs

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The International Rare Diseases Research Consortium (IRDiRC)

IRDiRC was created in 2011 to facilitate global multi-stakeholder collaboration in rare disease research. IRDiRC has set the following three goals for the decade 2017-2027

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Goal 1
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

Goal 2
1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options

Goal 3
Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patient

Inspirational goals which are still guiding the rare disease community, including ERDERA.

ERDERA is also hosting IRDiRC Scientific Secretariat.

https://irdirc.org

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Advancement of Treatments FOR RARE DISEASES

ERDERA: the European Partnership on Rare Diseases (2024-2031)

https://erdera.org/ ; https://cordis.europa.eu/project/id/101156595

ERDERA
European Rare Diseases Research Alliance

Objectives

to improve the health and well-being of the 30 million people living with a rare disease in Europe, by making Europe a world leader in RD research and innovation, to support concrete health benefits to rare disease patients, through **better prevention, diagnosis and treatment** and spearheading the digital transformational change in RD research and innovation (R&I).

- A **co-funded Partnership** (EU + Member States & Associated countries)
- Started **1st September 2024 f**or 7 years **– European Commission** press release
- **Hydrid nature:** joint calls (RD funding) + in-house activities (Clinical Research Network) to enhance the whole RD ecosystem
- Legacy of EJP RD co-fund, but also of other EU-funded projects such as Solve-RD, with ERNs highly involved !

380 M€ foreseen total budget
Up to 150 M€ EU contribution

183 partners
40 research funders
+ 81 research performing organizations
9 patients' organisations
3 EU infrastructures
23 others **(**univ**.,** hospitals, non-profit, public administration)
22 private-for-profit partners (industry & SMEs)

36 countries
26 EU Member States,
8 Associated countries
3 non-EU

ERDERA: A flagship co-fund with EU Member States and beyond !

Second part of the budget to be allocated based on further national commitments: stay committed !

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The power of collaborative research

A few examples

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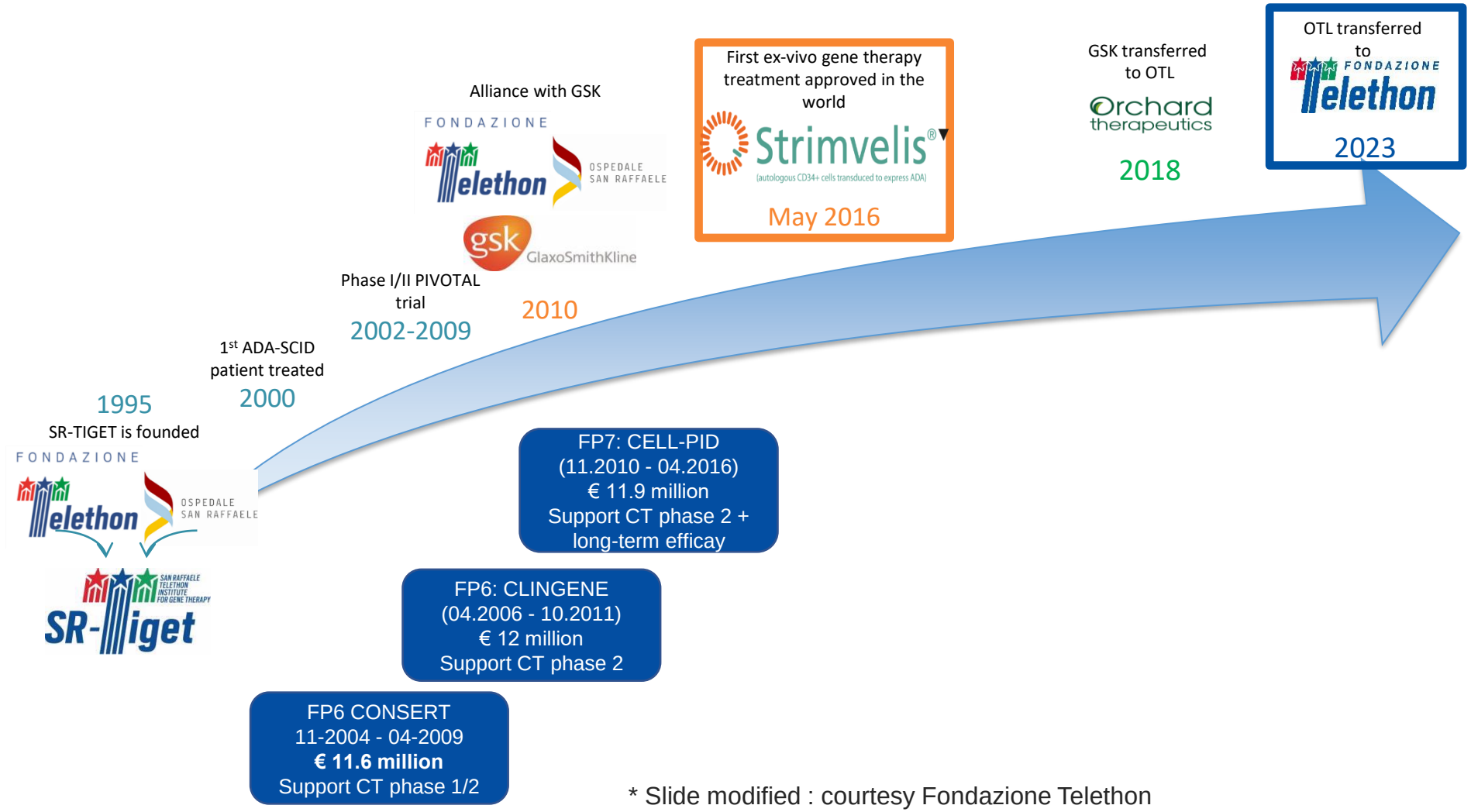
The story of Strimvelis, a gene therapy product used to treat severe combined immunodeficiency due to adenosine deaminase deficiency (ADA-SCID)



* Slide modified : courtesy Fondazione Telethon

Solve-RD: solving the unsolved Rare Diseases

Solve-RD
Solving the Unsolved Rare Diseases

Aim: understanding the (unknown) molecular etiologies of RD, and use genomic data to improve diagnosis

- Analysis of relevant genomics data of patients (almost **25k database**)
- Investigation of clinical diagnosis using omics (novel approaches to **capitalize NGS**)

- Potential to **decrease costs of healthcare** if correct diagnosis is promptly delivered

- Core collaboration of 4 ERNs, with outreach to all 24 ERNs. Now integrated in ERDERA Clinical Research Network, to include patients' data from more ERNs and also national undiagnosed programmes
- Outreach and ontological work to inform patients and clinicians

€15.3 million

2018-2024

26 partners

Multinational + multistakeholder consortium

17 universities
7 research organisations
1 public body
1 patients' organisation (EURORDIS)

10 countries
EU (DE, NL, FR, ES, CZ, BE, IT, P)
UK, US

European Commission

Advancement of Treatments FOR RARE DISEASES

HORIZON-HLTH-2022-DISEASE-06-04:
Development of new effective therapies for rare diseases

9 projects funded, ongoing

For a **total EU funding of 64,4 M€**

- EUropean network for neurodevelopmental RASopathies
- Gene Therapy for treatment of rare inherited Arrhythmogenic Cardiomyopathy
- Next-generation models and genetic therapies for rare neuromuscular diseases
- Nanomedicine Approach to Normalize Erythrocyte Maturation in Congenital Anemia by messenger RNA
- Novel advanced and repurposed therapeutics for vision restoration in a group of severe rare ocular surface diseases: from validation to first clinical investigations
- Accelerating drug repurposing for rare neurological, neurometabolic and neuromuscular disorders by exploiting SIMilarities in clinical and molecular PATHology
- Therapies for Renal Ciliopathies
- Drug REpurposing with Artificial intelligence for Muscular disorderS
- Light for double specificity and efficacy without burden

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But many other call topics support collaborative research projects on rare diseases…

Building a European innovation platform for the **repurposing of medicinal products (**HORIZON-HLTH-2021-DISEASE-04-02)

- REMEDi4All: Building a sustainable European Innovation Platform to enhance the repurposing of medicines for all
- REPO4EU: Precision drug REPurpOsing For EUrope and the world

Scaling up multi-party computation, data anonymisation techniques, and synthetic **data generation** (HORIZON-HLTH-2022-IND-13-02)

- SYNTHEMA: Synthetic generation of hematological data over federated computing frameworks (with ERN EuroBloodNet members)
- PHEMS: Pediatric Hospitals as European drivers for multi-party computation and synthetic data generation capabilities across clinical specialties and data types

Innovative tools for **use and re-use of health data** (in particular of electronic health records and/or patient registries) (HORIZON-HLTH-2021-TOOL-06-03)

- IDE4RC: Intelligent Ecosystem to improve the governance, the sharing and the re-use of health Data for Rare Cancers

And many more examples !

And we will continue funding….

Examples of call topics: generic, but relevant for rare diseases, in the Work Programme 2026-2027 of Horizon Europe:

2026 topics:

- HORIZON-HLTH-2026-02-DISEASE-12: European Partnership on Rare Diseases (ERDERA) (Phase 2)
- HORIZON-HLTH-2026-01-DISEASE-11: Understanding of sex and/or gender-specific mechanisms of cardiovascular diseases: determinants, risk factors and pathways
- HORIZON-HLTH-2026-01-CARE-01: Public procurement of innovative solutions for improving citizens' access to healthcare through integrated or personalised approaches
- HORIZON-HLTH-2026-01-TOOL-07: Establishing a European network of Centres of Excellence (CoEs) for Advanced Therapies Medicinal Products (ATMPs)

2027 topics:

- HORIZON-HLTH-2027-01-STAYHLTH-01: Addressing disabilities through the life course to support independent living and inclusion
- HORIZON-HLTH-2027-01-IND-01: Development of cell-free protein synthesis platforms for discovery and/or production of biologicals

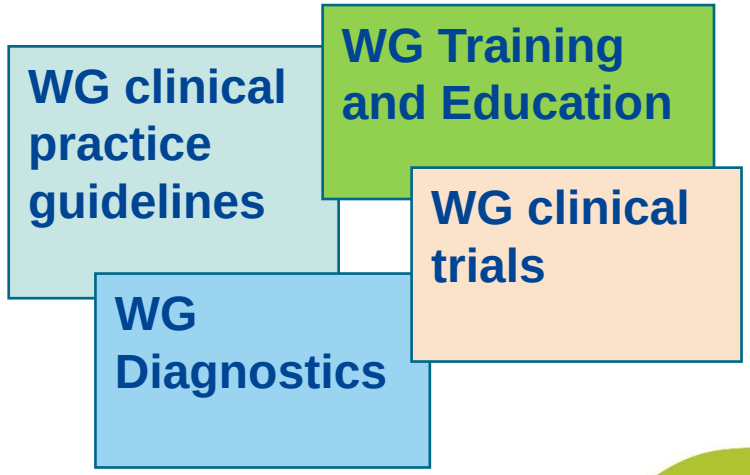
EU Cancer Mission - Rare Cancer Forum

Challenges for (very) rare cancer patients

- **Rare but not rare**: ~24% of all cancers are (very) rare*
- **Access**: few medicines (market, pipeline), few clinical trials and specialised centres, fragmented care pathways, limited industry interest
- **Inequality**: long time to diagnosis, few treatment options, lower 5-y overall survival
- **EU-added value:** accrual speed, statistics, funding, policy

What it is about

- R&I-centred initiative to facilitate structured dialogue and collaboration
- Identify and/or refine solutions in research and care to define strategic R&I priorities through coordinated action at EU and national levels
- Led by the Cancer Mission Board and five cancer societies (EHA, EORTC, ESMO, ESSO, SIOPE)
- Four working groups (for the moment)



*For definition and incidence of rare cancers see ESMO and RARECARE

Public-private projects under our Joint Undertakings

A few examples

in the field of rare diseases

Public Private Partnership with EFPIA

imi innovative medicines initiative

2008-2020

Public Private Partnership with Health Industries

innovative health initiative

2021-2027

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C4C (conect4children): Collaborative network for European clinical trials for children (2018-2025)

Objectives

- Create a sustainable, integrated pan-European collaborative paediatric network that will speed up of high-quality clinical trials in children, ensuring that the voices of young patients and their families are heard.
- Build the capacity for conducting multinational paediatric clinical trials for all disease areas and all phases of the clinical drug development process.

Outcomes

- C4C established an “Always on” paediatric clinical research network of > 400 experts, > 220 sites organized in 18 National Hubs
- Set up and deployed a service to integrate input from children, young people, and families into Expert Advice about on paediatric clinical research
- Training Health Care Professionals how to work with children and young people
- Established the European Young People’s Advisory Group Network (eYPAGnet)

https://conect4children.org/

154.4 M€ budget
67 M€ EU contribution
87.4 M€ EFPIA contribution

10 industry partners
34 universities, research organisations, non-profit private
1 SME
1 patient organisations (Eurordis)
10 third parties

20 countries
18 EU Member States
2 non-EU

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RealiseD: Comprehensive methodological and operational approach to clinical trials in rare and ultra-rare diseases (2025-2029)

Objectives

- Change the paradigm for clinical trials in ultra-rare diseases. By bringing together all stakeholders, including clinicians, methodologists, pharmaceutical industry, patients, regulators and HTA bodies.
- Catalyse the development and acceptance of innovative approaches for designing trials, (re-)using, analysing and interpreting data.
- Generate patient referral strategies to boost enrolment in future ultra-rare disease trials, and will set up a certification system to identify clinical trial sites capable of running studies on ultra-rare diseases.
- Work with several European Reference Networks (ERNs), via use cases from the fields of paediatric epilepsy, bone disorders, eye disease, and haematology and will be tested within ERDERA.

https://realised-ihi.eu/

17.4 M€ budget
8,5 M€ EU contribution
8,9 M€ Industry contribution

24 industry partners
22 universities, research organisations, non-profit private
3 SMEs
1 patient organisations (Eurordis)
2 third parties
EMA

13 countries
9 EU Member States
4 non-EU

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What have we learnt?

- No single country, discipline, or institution alone can tackle rare diseases
- Patient centricity is to address real-world needs
- The multi-disciplinary, multi-national, multi-stakeholders approaches match the complexity of the field
- One funding model does not fit-all
- Early advice and interaction with regulators is critical
- Commitment to goals are needed to serve scientific excellence

Innovations in rare diseases rarely remain rare

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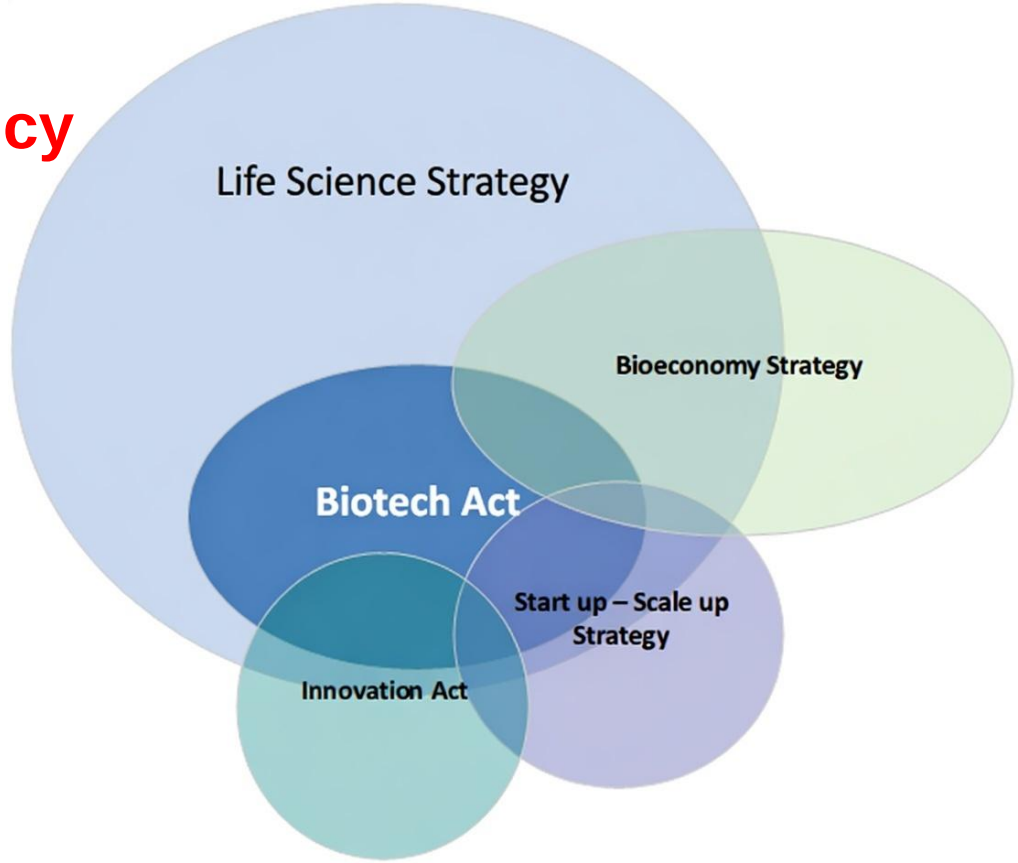
Conclusion:

a dynamic policy context



*Following this health-focused initiative, the Commission will address in 2026 the wider biotech ecosystem beyond health

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Strategy for European Life Sciences

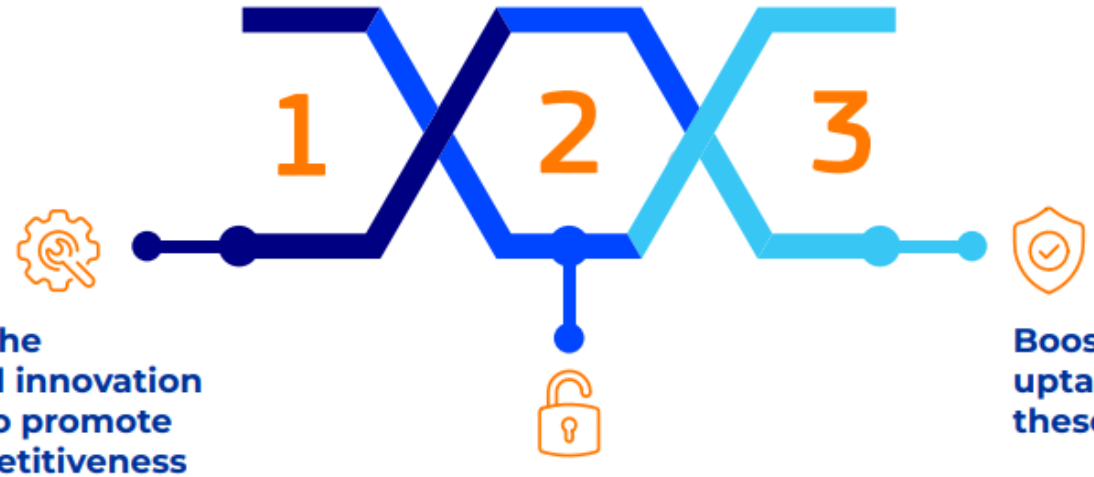
Adopted on 2 July 2025, the new strategy aims to **make Europe the most attractive place in the world for life sciences by 2030.**

Life sciences – the study of living systems, from cells to ecosystems – are **central to our health, environment and economy**.

A new strategy for European life sciences

Objective: to position the EU as the world's most attractive place for life sciences by 2030

3-phase approach to drive life science innovation:



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The European Biotech Act at a glance

Goals

- Boost innovation
- Support biotech sector
- Accelerate time to market

Structure

1) Industrial enablers

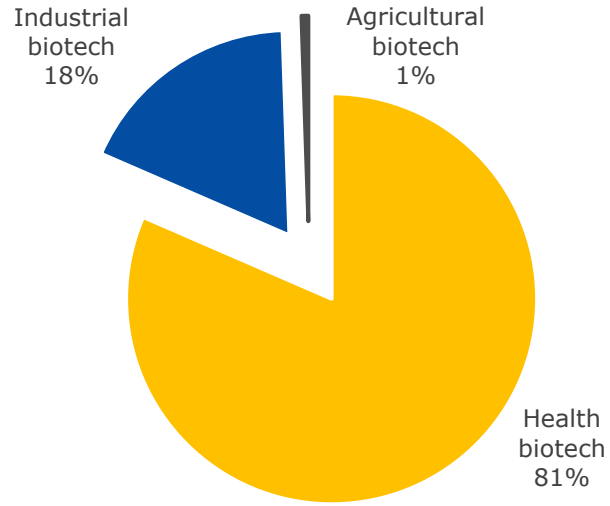
2) Regulatory simplification

Timeline

Commission proposal adopted on 16 Dec 2025

Scope

Focus on health (and food) biotech



Value added by Biotech sector in the EU, 2022

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Thank you for your attention !

More information ou EU research & innovation on rare diseases:

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

Cordis (database of EU-funded research projects): https://cordis.europa.eu/

IRDiRC: https://irdirc.org

ERDERA: https://erdera.org ; https://cordis.europa.eu/project/id/101156595

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