

Advancement of Treatments FOR RARE DISEASES

Witnessing and
Implementing Change over
the Past Two Decades -
The European
Commission's Action on
Rare Diseases and Therapy
Development



Organisers:



Funded by
the European Union

The conference is funded by the
European Union (project no 101297283)

Advancement of Treatments FOR RARE DISEASES

- Rare diseases remain one of the greatest challenges in modern medicine
- Progress over the past 20 years has transformed the landscape
- Europe's response has been built on solidarity, innovation, and cooperation
- The work ahead remains critical

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Advancement of Treatments FOR RARE DISEASES

Twenty Years Ago

- Long delays in diagnosis
- Fragmented expertise across Europe
- Limited research and treatment options
- Rare diseases often invisible in policy and healthcare systems
- Families faced uncertainty and isolation



The EU Response

- 2000: Adoption of the Orphan Medicinal Products Regulation
- A landmark statement of European values
- Incentives for innovation in rare diseases
- Support for research, scientific advice, and market exclusivity



Results Achieved



- From fewer than 10 authorised treatments to more than 200 orphan medicines
- Improved survival and quality of life for patients
- Greater awareness and investment in rare diseases
- Scientific breakthroughs transforming patient care

Building a Rare Disease Ecosystem

- National rare disease strategies across Member States
- Development of centres of expertise
- Better care pathways and multidisciplinary care
- Stronger cross-border collaboration



European Reference Networks

- Connecting specialists across Europe
- Cross-border expertise for complex cases
- Faster diagnosis and improved treatment decisions
- Reducing inequalities in access to expertise
- European cooperation in action



Scientific Transformation



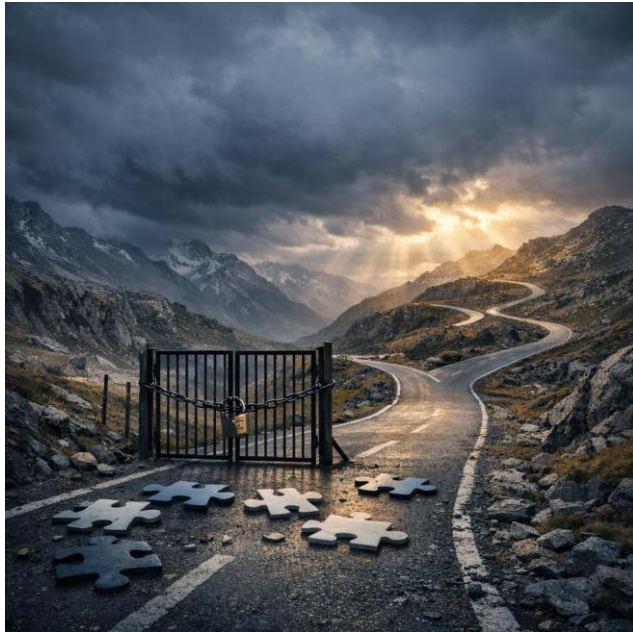
- Advances in genomics and personalised medicine
- Gene and cell therapies creating new possibilities
- RNA-based treatments and precision medicine
- AI and health data accelerating research
- Conditions once untreatable now entering clinical trials

The Role of Patients

- Patient organisations are now central partners
- Contributions to research priorities and trial design
- Stronger engagement with regulators and policymakers
- Policy grounded in lived patient experience



Remaining Challenges



- Unequal access to treatments across Europe
- Delays in pricing and reimbursement decisions
- High costs of advanced therapies
- Need for sustainable and fair healthcare systems
- Access must accompany innovation

The Future Agenda

- Reform of EU pharmaceutical legislation
- Faster access to medicines
- Better use of real-world evidence
- Earlier diagnosis through genomics
- Ethical and responsible use of health data
- Continued investment in centres of excellence



Closing Message



- Rare diseases are about people, families, and hope
- Europe has shown that change is possible
- Innovation must reach every patient
- A rare diagnosis must not mean a rare chance

“Hope is a diagnosis.
Hope is a treatment.
Hope is time.”