orphanet

25

1997 - 2022

Years of Orphanet

KNOW THE RARE

FOR BETTER CARE

RÉPUBLIQUE FRANÇAISE
Liberté
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Inserm
La science pour la santé
From science to health

Co-funded by the Health Programme of the European Union

elixir
Core Data Resource

IRDiRC
RECOGNIZED RESOURCES

THE HUMAN VARIOME PROJECT
RECOMMENDED SYSTEM

Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

25 Years of Orphanet

Orphanet was launched in 1997 by the INSERM (French National Institute for Health and Medical Research) to provide access to high-quality information on rare diseases. This initiative became a European endeavour from 2000, supported by grants from the European Commission: Orphanet has gradually grown to a Network of over countries, within Europe and across the globe.

To mark the 25th anniversary of Orphanet in 2022, we asked members of the wider rare disease community, our colleagues and our network members to share how Orphanet helps them to “know the rare” and how Orphanet has changed the rare disease ecosystem since its launch in 1997. Our network members and members of the Orphanet coordinating team also shared why they contribute to the Orphanet network.

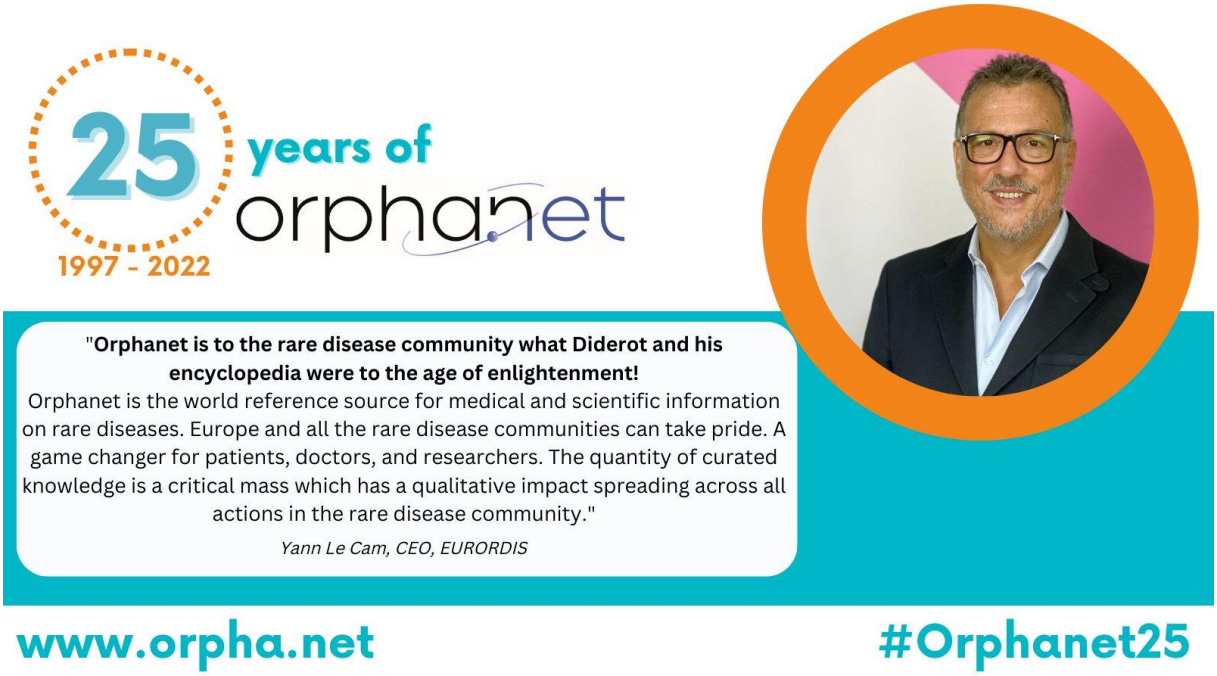
These contributions were shared on social media in late 2022, with the hashtag #Orphanet25, inviting the wider rare disease community to join the conversation and explain Orphanet’s impact in their work and lives.

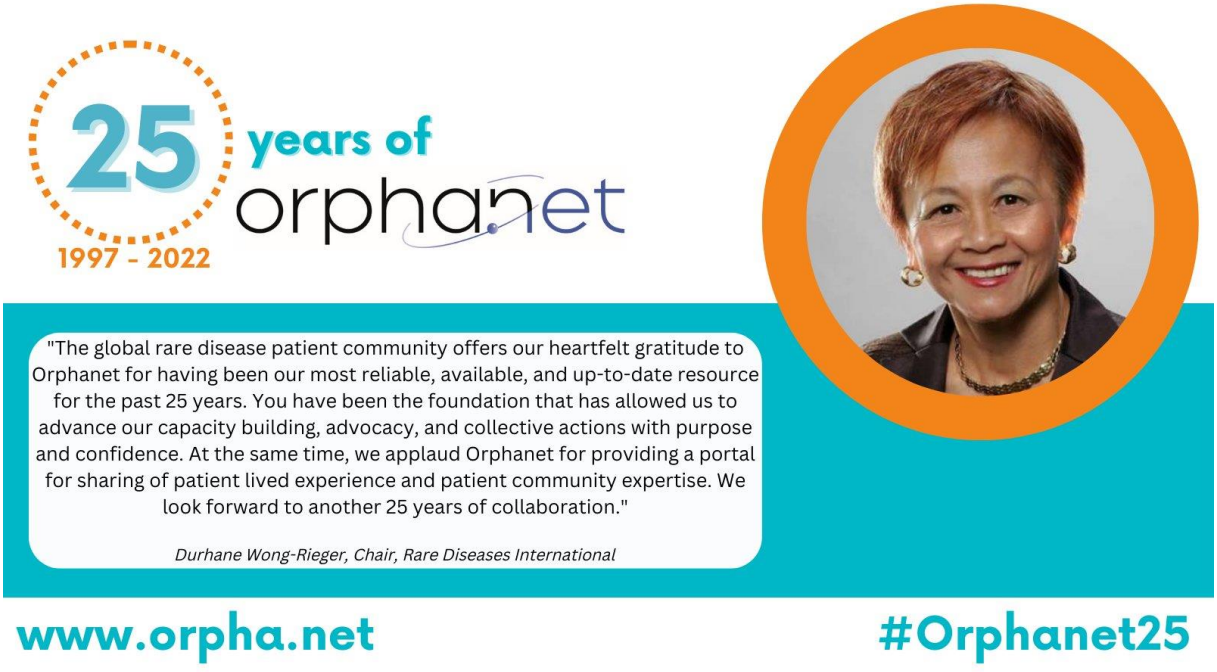
This booklet is a compilation of the input kindly provided from our colleagues across the world.

A word from our Director

Our impact for policy makers

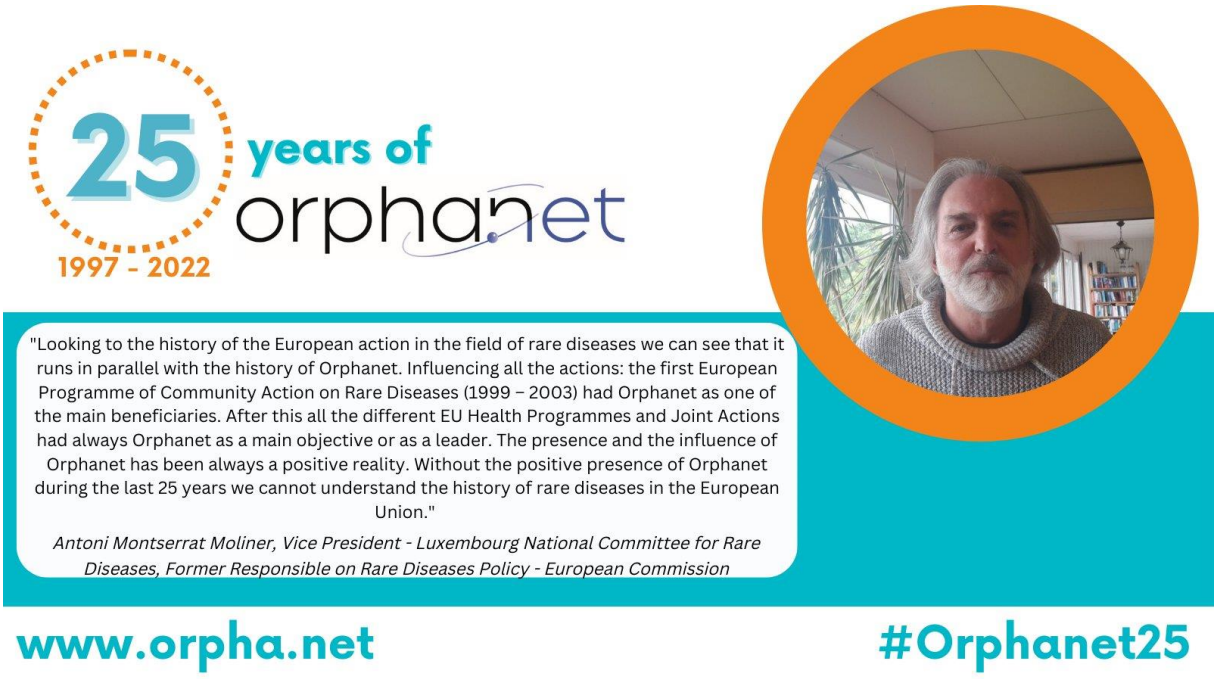
Our impact for patient advocates





Nos soutiens en France

Our impact on the RD ecosystem



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"We are proud to support Orphanet as an ELIXIR Core Data Resource, recognising its fundamental importance to the life science community and the need for its long-term preservation.
ELIXIR congratulates Orphanet on their 25th anniversary and look forward to working together on the new Genomic Data Infrastructure project providing a sustainable infrastructure for sensitive data."

Niklas Blomberg, ELIXIR Director

www.orpha.net #Orphanet25

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"By collecting, organising and sharing information and data on rare diseases, spanning from rare disease registries to research projects and professionals and by creating an ontology of rare diseases that is continuously updated and expanded, Orphanet has become an indispensable resource for patients, researchers, clinicians and for the whole rare disease community in Europe and globally."

Lucia Monaco, Past Chief Scientific Officer, Fondazione Telethon, Italy & Former Chair, International Rare Diseases Research Consortium (IRDiRC)

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"Orphanet serves as a knowledge hub for the European Rare Disease Community and beyond. In particular, the Orphanet Rare Disease Ontology has facilitated the standardisation of the clinical diagnosis terminology, preparing it for current and future applications, such as Artificial Intelligence. By collaborating and complementing with Orphanet we can reach our common goals faster and more effectively."

Sergi Beltran, Genome-Phenome Analysis Platform (GPAP), CNAG-CRG, Spain

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"Quelle chance de pouvoir disposer d'une telle ressource de connaissances et de documents/fiches pratiques sur des milliers de maladies rares !
Orphanet est un maillon essentiel de référence sur les maladies rares pour tout professionnel de santé mais aussi pour toutes les associations de malades à travers l'Europe !"

Marie-Pierre Bichet, Présidente de Maladies Rares info Services

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Further words on why they work with Orphanet:

“In my past work at Fondazione Telethon (the Italian charity funding and developing research on rare genetic diseases), I considered Orphanet as a key reference for the design of the database of Telethon-funded projects. In fact, all of them have been and are being annotated with the appropriate Orpha codes and are visible on the Orphanet database. The productive interaction with Orphanet allowed us to build a database centered on the rare diseases investigated in the research projects, represented in the biobanks and registries, addressed in the clinical trials and studied by the researchers and clinicians we have supported through the years.

I have had the pleasure to work with Orphanet in several international projects, such as RD-Connect and Rare2030 and within the International Rare Diseases Research Consortium (IRDiRC). In all cases. Orphanet has provided essential tools and resources for the successful development of the initiatives.”

Lucia Monaco, Past Chief Scientific Officer, Fondazione Telethon, Italy and past Chair, International Rare Diseases Research Consortium, (IRDiRC)

“Because I like to work in a coordinated way with people using a similar approach, a similar understanding and similar will to build tools at the service of patients. I like to work with competent and motivated people.”

Antoni Montserrat Moliner
Vice President of the Luxembourg National Committee for Rare Diseases
Member of the Board of ALAN-Maladies Rares Luxembourg
Member of the Eurordis Working Group on Newborn Screening
Active Senior on Public Health for the European Commission
Former Responsible on Rare Diseases Policy in the European Commission

Our Orphanet Network members


25 years of orphanet
1997 - 2022
"Orphanet has helped build and shape the rare disease community in our country, in Europe, and even worldwide during these past 25 years in a unique way. Almost everything we have achieved in Austria for rare diseases we eventually owe to Orphanet."
Ursula Unterberger, Orphanet Austria, Medical University of Vienna
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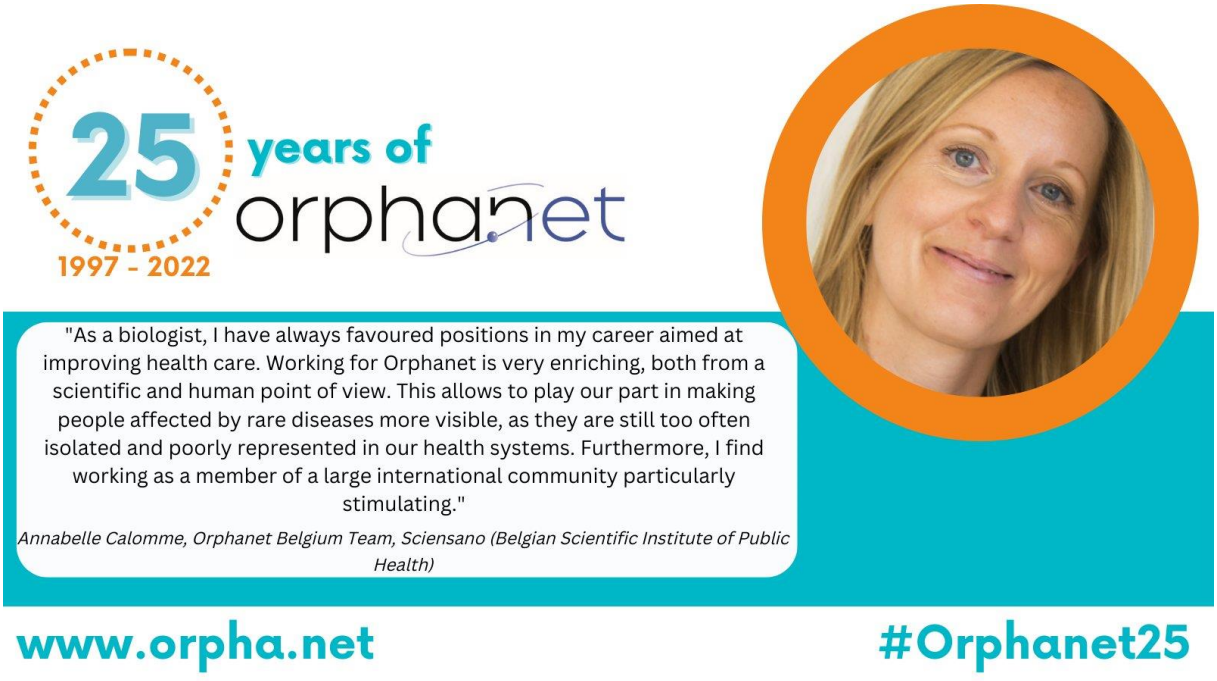
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"As a biologist, I have always favoured positions in my career aimed at improving health care. Working for Orphanet is very enriching, both from a scientific and human point of view. This allows to play our part in making people affected by rare diseases more visible, as they are still too often isolated and poorly represented in our health systems. Furthermore, I find working as a member of a large international community particularly stimulating."
Annabelle Calomme, Orphanet Belgium Team, Sciensano (Belgian Scientific Institute of Public Health)
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"It's very rewarding to see how we can help improve the resources available in the interest of achieving better care for people living with a rare disease."

Orphanet Spain Team, CIBERER

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"Families and health professionals address the Orphanet platform for getting accurate information on a specific rare disease including making the link with expert centers worldwide. In Israel, we are actually working on the translation in the local language to facilitate its use.
We think that connection worldwide is key for better care of the patients living with rare diseases. Orphanet is "The tool". We are deeply convinced that giving appropriate information is key to care for the patients. Networking is essential even more when you work and live in a small country."

Prof. Annick Rotschild, Orphanet Israel, Insitute of Rare Diseases, Sheba Medical Center

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Further words on why they work for Orphanet…

"Orphanet has connected healthcare providers, researchers and patients to each other and created an information and data portal for rare diseases and orphan drugs.

Orphanet is a welcoming community that wants to contribute and interact with all stakeholders in the field of rare diseases!"

Wendy A.G. van Zelst-Stams, Coordinator Orphanet Netherlands, Radboud University Medical Centre

"There is an obvious professional reason, however, even more importantly it is sincerely gratifying and pleasing to meet such great, dedicated team at Orphanet. i.e. of likely-minded experts who perform their excellent work truly with their hearts. I very much appreciate how the entire Orphanet team has achieved world-wide reputation even in the absence of adequate funding! Thus, more then ever they require our support – from national coordinators, since we are all „in" for our patients and their families."

Milan Macek Jnr, Coordinator Orphanet Czechia

Words from the Orphanet Coordinating Team

"I am proud to be part of such a useful, ambitions and exciting project with such clear European and International added-value. I am thankful to collaborate with passionate and dedicated people from all across the rare disease community to deliver data, information and resources that can help improve the lives of those living with a rare disease." *Charlotte Rodwell, Partnerships and Steategic Communications Officer, Orphanet Coordinating Team*

"Orphan drugs repository maintenance,

Rare diseases are rare, but rare disease patients are numerous,

Patient-centric resource,

High quality information and expertise in rare diseases,

Analyses of rare disease environnement,

Network of international, multicultural and multilingual communities sharing knowledge on rare disease,

Expert resources catalog development and exploitation,

Team work."

Julie Bruyère-Zrelli, Orphan Drugs Database Manager, Orphanet Coordinating Team

« Deux fois par mois, OrphaNews contribue, lentement mais sûrement, à relayer les actualités de l'environnement « maladie rare ». Savoir, c'est pouvoir… » *Henri Jautrou, Rédacteur OrphaNews France*