

# orphanet



Years of Orphanet



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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



"In the constantly evolving ecosystem of rare diseases, Orphanet is a tool, a partner, an actor, but also a common language to understand each other. Orphanet is above all a passionate team of committed professionals who work hand-in-hand with equally passionate and committed players in fields as diverse as health, research, data science... in order to co-build a better world for people who suffer from rare diseases.

We all share the same passion: making the rare visible.

I am proud and honored to serve this amazing 25 year-old young adventure."

*Ana Rath, Director of Orphanet, INSERM, France*

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# Our impact for policy makers



"Orphanet depuis 25 ans, ce sont des femmes et des hommes passeurs d'information, de formation et d'espoir. Leur dynamisme et leur rigueur aident la communauté des maladies rares à se rassembler, à se reconnaître pour avancer plus loin ensemble. Orphanet est la boussole pour beaucoup d'entre nous. Toujours présent, toujours disponible et attentif à construire une société plus juste et inclusive. Orphanet a permis de construire et structurer notre politique maladies rares en France. Toujours présente pour accompagner nos 3 plans nationaux maladies rares. Orphanet rassemble la communauté des maladies rares : les personnes ayant une maladie rare, les aidants, les associations, les médecins, les chercheurs et les Etats partout dans le monde."

*Anne-Sophie Lapointe, Cheffe de Projet mission maladies rares au Ministère de la santé et de la prévention*

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"Work on rare diseases is built on important shared values: solidarity, inclusion, equality, and human dignity. ORPHANET is an important partner for all our work related to rare diseases."

*Dr. Andrzej Rys, Director, DG SANTE, European Commission*

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# Our impact for patient advocates



**"Orphanet is to the rare disease community what Diderot and his encyclopedia were to the age of enlightenment!"**

Orphanet is the world reference source for medical and scientific information on rare diseases. Europe and all the rare disease communities can take pride. A game changer for patients, doctors, and researchers. The quantity of curated knowledge is a critical mass which has a qualitative impact spreading across all actions in the rare disease community."

*Yann Le Cam, CEO, EURORDIS*

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"The global rare disease patient community offers our heartfelt gratitude to Orphanet for having been our most reliable, available, and up-to-date resource for the past 25 years. You have been the foundation that has allowed us to advance our capacity building, advocacy, and collective actions with purpose and confidence. At the same time, we applaud Orphanet for providing a portal for sharing of patient lived experience and patient community expertise. We look forward to another 25 years of collaboration."

*Durhane Wong-Rieger, Chair, Rare Diseases International*

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# Nos soutiens en France



"L'information est le nerf de la guerre. Depuis 25 ans, Orphanet est un levier puissant pour faire exister les maladies rares et les sortir de l'oubli. En s'appuyant sur un important réseau international d'experts, Orphanet met à la disposition de tous -malades, médecins, chercheurs, industriels, administrations- une information dont l'exhaustivité est unique au monde. Orphanet a ainsi contribué à l'émergence de politiques publiques, en France mais également en Europe. Nous sommes très fiers d'avoir contribué à créer cet outil indispensable au combat contre les maladies rares que nous soutenons sans discontinuer depuis 25 ans grâce au Téléthon. Orphanet le montre : l'union fait la force ! "

*Laurence Tiannot-Herment, Présidente de l'AFM-Téléthon*

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"En favorisant un meilleur accès à la connaissance entre toutes les parties prenantes, Orphanet contribue à une meilleure démocratie sanitaire entre les populations et dans les territoires. En tant que Fondation d'un groupe d'assurance mutualiste, nous sommes très attachés à cette dimension d'égalité à laquelle Orphanet contribue. Et en tant qu'acteur privé de référence des maladies rares il est totalement évident pour la Fondation d'être partenaire de ce portail jamais égalé."

*Sophie Dancygier, Déléguée générale, Fondation Groupama*

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# Our impact on the RD ecosystem



"Looking to the history of the European action in the field of rare diseases we can see that it runs in parallel with the history of Orphanet. Influencing all the actions: the first European Programme of Community Action on Rare Diseases (1999 - 2003) had Orphanet as one of the main beneficiaries. After this all the different EU Health Programmes and Joint Actions had always Orphanet as a main objective or as a leader. The presence and the influence of Orphanet has been always a positive reality. Without the positive presence of Orphanet during the last 25 years we cannot understand the history of rare diseases in the European Union."

*Antoni Montserrat Moliner, Vice President - Luxembourg National Committee for Rare Diseases, Former Responsible on Rare Diseases Policy - European Commission*

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"If you take rare diseases seriously you cannot NOT collaborate with Orphanet. It is a key partner! Working without Orphanet is like trying to read without glasses when you need them: you attempt to decrypt something, but everything is blurry and makes little sense."

*Daria Julkowska, EJP-RD Coordinator, IRDiRC*

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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

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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



"Orphanet is a crucial "home-base" for all medical specialties dealing with rare diseases. Without Orphanet the field of rare diseases would not have achieved such a remarkable progress, culminating thus far in the successful establishment of pan-European ERN networks. "

Milan Macek Jr, Orphanet Czechia, Charles University Prague

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"Rare diseases have problems everywhere. In Finland, a country with a small population (5,5 M), long distances and small language group add to the problems. So collaboration with Orphanet is extremely important for us."

Helena Kääriäinen, Orphanet Finland, Finnish Institute for Health and Welfare

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"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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"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 than 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



"What is rare is precious.  
For 25 years now Orphanet has been a core resource in the rare diseases field. It's also a journey, a network of which I'm sincerely proud to be part. There has been many years of technical challenges, new projects and services, millions of users across the world and above all the will to serve the common good and the rare diseases community."

*Marc Hanauer, CTO & Deputy Director of Orphanet, INSERM, France*

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"I am proud to be part of such a useful, ambitious 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 Strategic 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 environment,**

**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*