

'Rare is not rare'

New scientific paper confirms 300 million people living with a rare disease worldwide

Paris, 17 September 2019 - A new scientific paper **confirms the number of people living with a rare disease is estimated at 300 million worldwide**. [The open-access article](#), written by Orphanet (coordinated by INSERM, the French National Institute of Health and Medical Research), EURORDIS-Rare Diseases Europe and Orphanet Ireland (based at the National Rare Diseases Office, Mater Misericordiae University Hospital, Dublin, Ireland), [is today published in the European Journal of Human Genetics](#).

According to the research published in the paper, rare diseases currently affect at any point in time **3.5% - 5.9% of the worldwide population**, equivalent to a conservative estimate of **300 million people worldwide** (4% of the an estimated world population of 7.5 billion¹), the number used until now by EURORDIS and Rare Diseases International. The analysis shows that there are **over 6,000 clinically defined rare diseases²**, **72% are genetic**, and of those **70% of rare diseases start in childhood**.

This is the first time that a robust analysis of the global prevalence of rare diseases has been conducted using the publicly available epidemiological data in the Orphanet (www.orpha.net) database. The figures presented in the paper are derived from data from 67.6% of the prevalent rare diseases. As this analysis does not take into account rare cancers, nor rare diseases caused by rare bacterial or viral infectious diseases and poisonings, the **number of people affected by rare diseases is likely considerably higher**.

Ana Rath, co-author of the paper, Director of Orphanet, stated, "Even if these are the best figures we can obtain today, they likely underestimate the number of rare disease patients still not visible in healthcare and social care systems. Having a specific codification system for them in national systems will help obtain definitive figures and, more importantly, produce data needed to adapt healthcare systems".

Yann Le Cam, co-author of the paper, Chief Executive Officer of EURORDIS and member of the Council of Rare Diseases International, commented, "Collectively rare diseases are not rare. The findings published in this paper support years of efforts from the rare disease community to advocate for the prioritisation of rare diseases as a public health priority that affects millions of people around the world, not just the few. No one can deny the significance of the global rare disease population anymore, a critical mass of people who have been the health orphans so far, with acute needs for

¹ Nguengang Wakap S et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics*, 2019. Worldwide population in 2017 of an estimated 7 550 000 000 people.

² In Europe, a disease is defined as rare when it affects no more than 1 in 2,000 people, as per the [Regulation \(EC\) N°141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products. 2000](#).

healthcare, access to innovative treatments and a social system that supports their right to reach their highest potential of well-being”.

Findings presented in the paper, based on the analysis of the 67.6% of the prevalent diseases, include that:

- 149 rare diseases analysed affect around 80% of the rare disease population. These diseases are the most prevalent among the diseases, with each affecting **100 – 500 people per 1 million**.
- The next most prevalent 241 rare diseases analysed affect **10 – 100 people per 1 million people**.
- Collectively, these nearly 400 diseases affect at least 98% of the rare disease population with a prevalence of **10 – 500 people per 1 million people**. They can be grouped by therapeutic areas and care services built around these therapeutic areas.
- The remaining diseases analysed are the less prevalent diseases as they affect **less than 10 people per 1 million people**.

Building on these important data, the analysis will be developed in the future to cover rare diseases caused by infectious agents, environmental factors, bacteria or viruses, poisoning and rare cancers; and to deepen the research for around 24 rare disease groupings (mirroring the grouping adopted for the European Reference Networks), all with the aim to ultimately help inform health and social care planning for people living with a rare disease and their families and to streamline research on rare diseases. Future registry research and the implementation of rare disease codification in healthcare systems will further refine the estimates.

Read the publication:

Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database
<https://www.nature.com/articles/s41431-019-0508-0>

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About Orphanet – INSERM

[Orphanet](#) is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet also maintains the Orphanet rare disease nomenclature (ORPHA number), essential in improving the visibility of rare diseases in health and research information systems. The Orphanet Network, 39 countries across the world contributing to the aforementioned goals, is coordinated by [INSERM](#).

The integral role played by Orphanet in the research and care spheres has led to its recognition as an [IRDIRC Recognised Resource](#), and integration in the French node of [ELIXIR](#), a European research infrastructure uniting Europe's leading life science organisations. Orphadata, Orphanet's data download platform, and the Orphanet Rare Disease Ontology, are [ELIXIR Core Data Resources](#).

About EURORDIS-Rare Diseases Europe

[EURORDIS-Rare Diseases Europe](#) is a unique, non-profit alliance of over 800 rare disease patient organisations from over 70 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

About the National Rare Diseases Office of Ireland

The National Rare Diseases Office provides current and reliable information about genetic and rare diseases to patients, families and healthcare professionals in Ireland. The office was established in 2015 by the Irish Health Service Executive and is staffed by healthcare professionals who have significant experience working with people affected with rare diseases. The office identifies and catalogues Irish rare disease resources on Orphanet; provide evidence based information services; and build links with ERNs. The website (<http://www.rarediseases.ie/>) provides information about national social care supports, educational resources and eLearning modules.