

Kirsten Axelsen<sup>a</sup>, Annie Kennedy<sup>b</sup>, Lisa Feng<sup>c</sup>, Charlotte Poon<sup>d</sup>  
<sup>a</sup>Charles River Associates, New York, NY, USA <sup>b</sup>EveryLife Foundation for Rare Diseases, Washington, DC, USA <sup>c</sup>Alexion, AstraZeneca Rare Disease, Boston, MA, USA  
<sup>d</sup>Charles River Associates, London, United Kingdom

For more information, please contact Charlotte Poon at [cpoon@crai.com](mailto:cpoon@crai.com)



## Introduction

- 30 million people** in Europe live with an estimated **7,000 rare diseases** (RDs), collectively representing a significant public health issue<sup>1,2</sup>
- Even so, we **know little** of the impact of RDs experienced by patients, their families and the economy
- Objectives:** To address this information gap, we estimate the **economic and social impact** for persons living with rare diseases (PLWRD) for 43 RDs across 9 European countries

FR

DE

IT

NL

PL

RO

ES

SE

UK

## Methods

- Reviewed the **literature** on the economic cost of RDs
- Developed a **patient survey** with the methodology informed by our **literature review** and **expert input** from 45 RD specialists
- Disseminated the survey between February and May 2024 and collected **detailed information** about the diagnostic journey, healthcare utilisation, treatments and the wider impact associated with living with a RD from July to December 2023
- Quantified the **economic cost of 43 RDs** using a **prevalence-based approach**

Sample

1,317

Total respondents

545

PLWRD

772

Reference group (without a RD)

## Results

### PLWRD incurred higher costs to society

**Total excess costs**

Direct medical costs	Direct non-medical costs	Indirect costs
<b>€184.5B</b> Includes physician and other HCP visits, procedures, Rx medications, inpatient days at hospitals, telehealth and home health appointments and durable medical equipment.	<b>€37.4B</b> Includes paid caregivers, transportation and accommodation for health care, disease-related daily living arrangements, long-term care facility days and visits to various therapists.	<b>€27.5B</b> Includes lost productivity costs from absenteeism, presenteeism, early retirement and a forced shift to part-time work endured by the person living with the RD and their caregivers.

**~ €249.3billion per year**

**Annual per-capita excess cost**

<b>€20,200</b> Reference group annual per-capita cost	<b>€121,900</b> RD group annual per-capita cost	<b>6X</b> Higher
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### PLWRD experience inequitable care

25%

Of PLWRD were **misdiagnosed** at least once in their diagnostic journeys

32%

**Lower health-related quality of life** reported by PLWRDs compared to reference group

3X

**Longer time to diagnosis if misdiagnosed** compared to those without a misdiagnosis (36 vs 11 months)

78 days

**Of lost productivity per year** for each person with a rare disease across themselves and their caregivers

4X

**Faster diagnosis** when targeted EMA-approved treatments were available at symptom onset

1 year

**Longer time to diagnosis for women** compared to men, and **+1.5 year longer** time to start treatment

## Conclusions

### Our findings

- The economic impact of living with a RD extends beyond healthcare costs. A significant portion of the cost reflects the **reduced earnings, productivity and career opportunities** of PLWRD and their caregivers
- RD patients face a lengthy diagnostic journey, with each **misdiagnosis adding costs and negatively impacting the quality of life** for PLWRD and their caregivers
- Accessing specialist care and treatment** are key drivers affecting non-medical and indirect costs and improving quality of life for patients and caregivers

### Policy implications

- Public policies on **RD innovation and treatment access** should consider the **full breadth** of impact on PLWRD, caregivers and society
- Policies affecting **newborn screening, genome sequencing, and other diagnostic tools** need to be prioritised to **reduce time to diagnoses**
- Improving **treatment equity and physician awareness** can speed up access to treatment, helping **reduce both medical and non-medical costs**

**References**  
1. Rare diseases - European Commission. March 8, 2024. Accessed May 8, 2024. [https://health.ec.europa.eu/european-reference-networks/rare-diseases\\_en](https://health.ec.europa.eu/european-reference-networks/rare-diseases_en)  
2. Addressing the challenges of persons living with a rare disease and their families: resolution /:adopted by the General Assembly. Published online December 16, 2021. Accessed May 14, 2024. <https://digitalibrary.un.org/record/3953832>

**Disclosures**  
K. Axelsen, C. Poon are employees of Charles River Associates, which was hired by Alexion, AstraZeneca Rare Disease to provide research/writing support for this study. Charles River Associates is an economic consultancy company with a long-established reputation for independent analysis. The views expressed herein are the views and opinions of the authors and do not reflect or represent the views of Charles River Associates or any of the organisations with which the authors are affiliated. A. Kennedy is an employee of EveryLife Foundation for Rare Diseases and L. Feng is an employee of Alexion, AstraZeneca Rare Disease who may own stock and/or hold stock options in the Company.