The Economic Cost of Living with a Rare Disease Across Europe

Overview of research

November 2024

ALEXION®
AstraZeneca Rare Disease

CRA Charles River Associates

Today's Agenda

1. Review the objectives of the research
2. Review the methods and approach
3. Present the key study findings
4. Q&A

Our study addresses a knowledge gap on the burden of rare diseases in Europe, and builds on the landmark study* from ELF

01 Although there has been significant progress on awareness regarding rare diseases, there are still major barriers to rare disease care

02 There is a significant knowledge gap, there are only economic cost studies specific to a single RD or a specific European country

03 Understanding the burden of disease would inform the need for continued prioritization, particularly given the shifting rare disease policy landscape in Europe with the General Pharmaceutical Legislation

*In 2021, EveryLife Foundation for Rare Diseases published The National Economic Burden of Rare Disease Study, providing a comprehensive assessment of the total economic burden of RDs in the US in 2019 (link to report)

Our research is unique in collecting data through a patient survey to estimate the economic cost of rare diseases in 9 European countries

Purpose

To quantify the economic impact of rare diseases in Europe using a patient survey approach to support the policy prioritization of rare disease innovation, treatment, and access.

Establish this evidence in collaborative partnerships with leading patient advocates globally for credibility and dissemination.

Specific Objectives

To describe the social and economic impact of rare diseases compared to reference group.

Evaluate the direct medical, direct non-medical, and indirect costs incurred by PLWRDs and their caregivers in Europe.

To better inform policy decisions that affect awareness, diagnosis, treatment, and access.

Geographic Scope

Rare Diseases

43 diseases: 29/43
(67%) are genetic diseases

8 DISEASE CATEGORIES: (N=545)

- neurologic (n=8)
- hematology (n=5)
- immunology (n=9)
- pulmonary (n=3)
- congenital malformations (n=8)
- endocrine (n=3)
- oncology (n=2)
- metabolic (n=5)

Compared to reference group (general population without rare disease)

We used a 5-step approach to understand previous studies, design a survey, then develop policy implications based on our findings

Study Approach

Activities

1. KO + Alignment	2. Literature Review	3. Survey + Interviews	4. Data Analysis	5. Report Writing
• Established variables of interest • Outreach to partners, established modes of working together	• Conduct secondary research, including review of published burden studies	• Validation interviews with KOLs • Design patient survey • Field survey	• Quantification of costs • Develop conclusions and implications	• Summarise findings into a paper and iterate

CRA Charles River Associates

We had close collaboration throughout the study with experts in RD space

Built on relationships Alexion has established globally

Expert Contributors

The approach

- Shared input on study design
- Reviewed the analysis plan and interim results
- Reviewed final results and report
- Shared input on presentation of report and dissemination ideas

We derived costs from direct medical, direct non-medical and indirect categories and included QoL as a nonmonetary cost

Elements	Overview			
Design	**Direct Medical**	**Direct Non-Medical**	**Indirect**	**Non-monetary**
	• Outpatient hospital visits and procedures • Inpatient stays and procedures • Outpatient clinic visits and procedures • Medicines • Durable medical equipment (DME) • ER and ambulance • Patient and caregiver mental health • GP visit • Home visit • Telehealth	• Cost of caregivers • Various therapists • Disease-related arrangements • Long-term care facility • Transportation and accommodation	• Absenteeism • Presenteeism • Early retirement • Loss from transition to part-time	• Patient and caregiver QoL

The more ‘common’ rare diseases were selected and validated by KOLs as relevant diseases seen in clinics

Rare Diseases							
Congenital/ chromosomal	**Hematology**	**Immunology**	**Oncology**	**Endocrine**	**Metabolic**	**Neurologic**	**Pulmonary**
Angelman syndrome	Acquired aplastic anemia	Juvenile idiopathic arthritis	Histiocytosis	Acute intermittent porphyria	Fabry disease	Amyotrophic lateral sclerosis	Cystic fibrosis
Deletion 5p	Atypical hemolytic uremic syndrome	Pemphigus vulgaris	Multiple myeloma	HTTR Amyloidosis	Gaucher disease	Ataxia telangiectasia	Idiopathic pulmonary fibrosis
Epidermolysis bullosa	Beta thalassemia major	Scleroderma		Phenylketonuria	Hunter syndrome (Mucopolysaccharidosis II)	Autoimmune encephalitis	Pulmonary arterial hypertension
Fragile X syndrome	Haemophilia	Common variable immune deficiency			Mucopolysaccharidosis	Christianson syndrome	
Ornithine transcarbamylase deficiency	Sickle cell disease	Hereditary angioedema			Danon Disease	Duchenne muscular dystrophy	
Prader-Willi syndrome		Aspergillosis				Early onset familial Alzheimer’s disease	
DiGeorge syndrome		ANCA-associated vasculitis				Myasthenia gravis	
Leber neuropathy						Spinal Muscular Atrophy	

The list includes a mix of disease against the following criteria:

- Age of onset
- On NBS panel
- Availability of EMA approved treatment
- Treatment has orphan designation

Key Study Findings: we concluded that PLWRD and their caregivers experienced a significant economic cost

Total Excess Costs (on top of reference group)

Direct Medical Costs		Direct Non-Medical Costs		Indirect Costs
€185B	+	€38B	+	€28B

~ €250 billion per year

Across 43 rare diseases in 9 European countries

Annual Per-Capita Excess Cost

€20,200
Reference Group Annual Per-Capita Cost

€121,900
RD Group Annual Per-Capita Cost

→ **6X Higher**

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 **treatment start**

CRA Charles River Associates

Key conclusions and implications

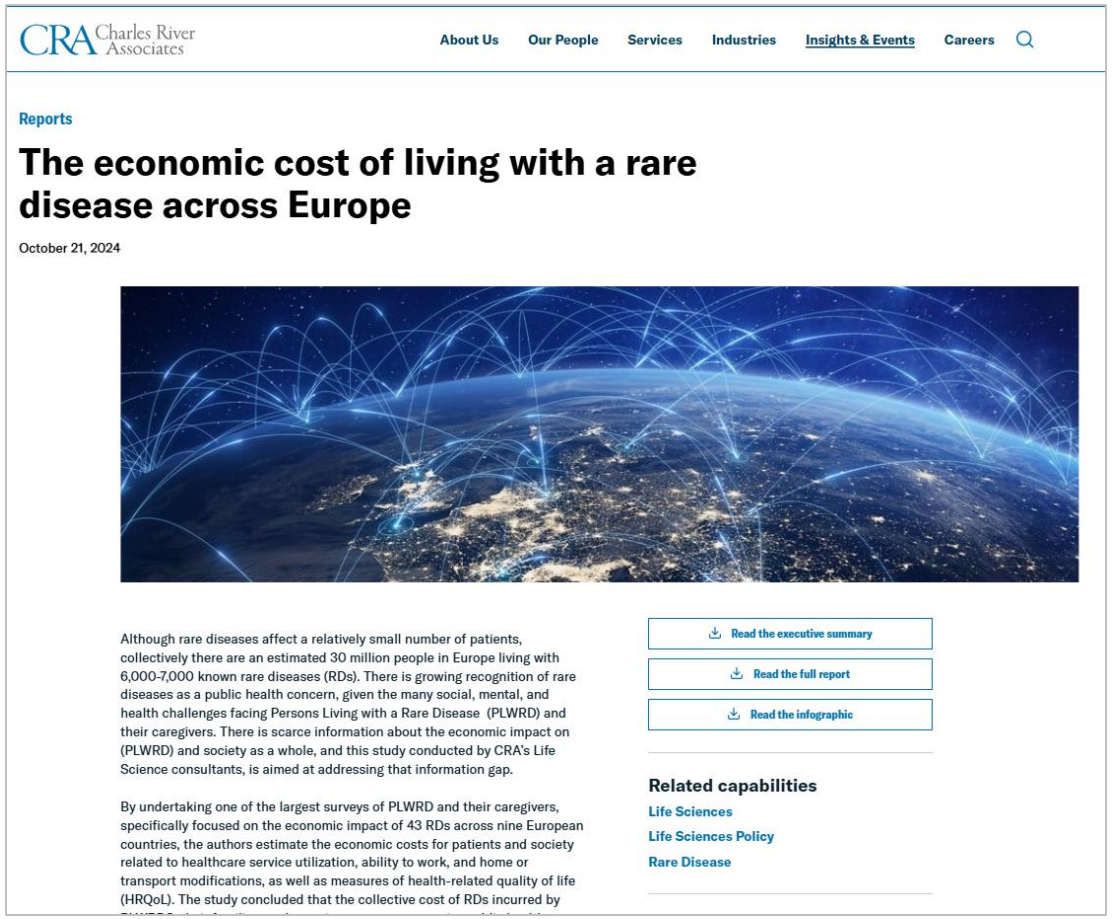
Our findings

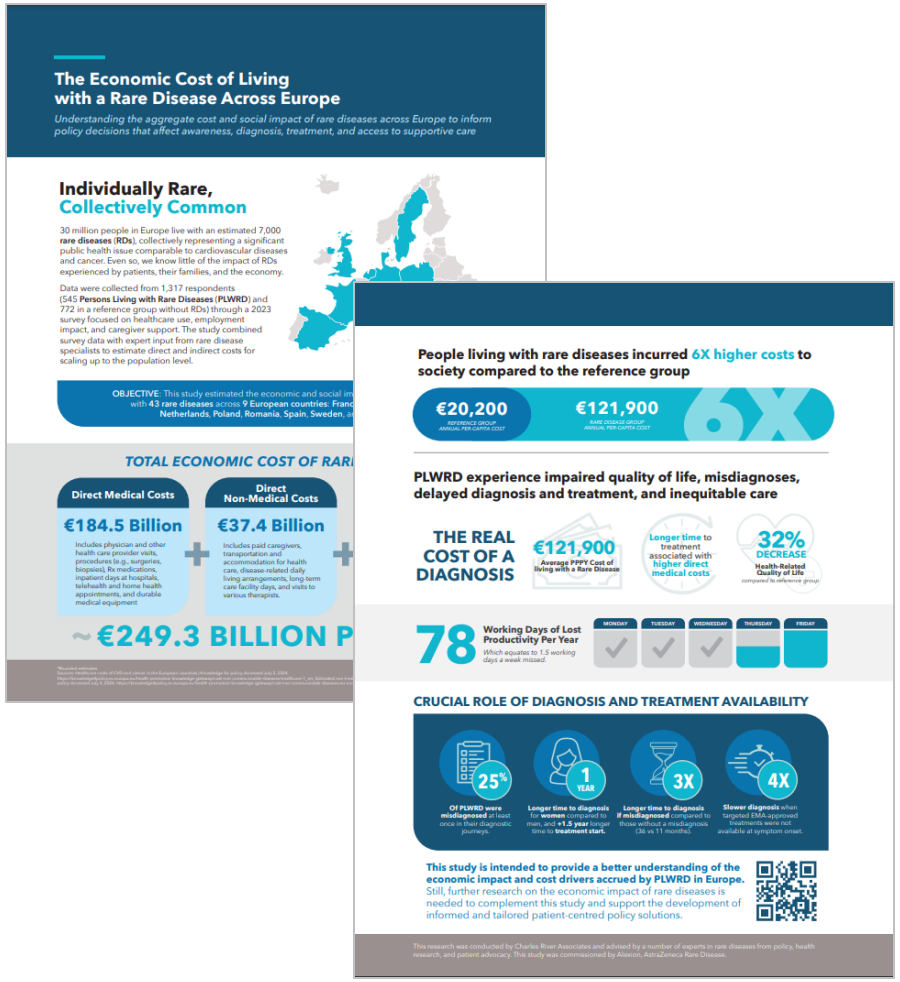
1. The economic impact of living with a RD extends beyond healthcare costs. A significant portion of the cost reflects **reduced earnings, productivity, and career opportunities** of PLWRD and their caregivers
2. RD patients face a lengthy diagnostic journey, with each **misdiagnosis adding costs and negatively impacting the quality of life** for PLWRD and their caregivers
3. **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

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

The report is hosted on CRA's website and we have developed a 2- page infographic to support dissemination





CRA Charles River Associates

We are now conducting a similar study in Japan with the list of rare disease and analysis plan tailored to local needs

Rare Diseases have been identified through the Nanbyo (Intractable Diseases) List

ASrid

Advocacy Service for Rare and Intractable Diseases

特定医療費（指定難病）受給者証所持者数，年齢階級・対象疾患別

(出典：令和4年度衛生行政報告例(令和4年度末現在))

表示番号	総数	総数	0～9	10～19	20～29	30～39	40～49	50～59	60～69	70～74	75歳以
6	パーキンソン病(ICD10:G20)	143,267	–	4	17	138	1,109	5,598	19,866	26,688	89,847
97	潰瘍性大腸炎	141,387	4	1,150	15,089	21,816	30,396	30,318	21,285	9,610	11,7
49	全身性エリテマトーデス	65,145	–	222	4,833	8,746	14,408	14,923	10,529	5,211	6,
96	クローン病	50,184	4	613	8,948	11,054	12,404	9,818	4,369	1,405	1
69	後縦靭帯骨化症	31,571	–	–	27	255	1,728	4,554	6,727	5,511	12
51	全身性強皮症	27,013	–	11	189	531	1,754	4,188	6,551	5,152	8
18	脊髄小脳変性症(多系統萎縮症を除く。)	26,476	–	30	315	895	1,952	3,555	5,309	4,491	9,
11	重症筋無力症	26,387	3	96	559	1,205	2,659	4,418	5,385	3,840	8,2
50	皮膚筋炎／多発性筋炎	26,046	1	36	386	1,046	2,884	5,328	6,388	4,068	5,90
13	多発性硬化症／視神経脊髄炎	23,105	1	97	1,306	3,161	5,927	6,071	3,454	1,435	1,653
306	好酸球性副鼻腔炎	22,340	–	24	517	2,033	4,730	6,707	5,099	1,879	1,351
90	網膜色素変性症	21,263	5	79	265	631	1,589	2,832	4,110	3,529	8,223
78	下垂体前葉機能低下症	19,693	12	519	1,915	2,138	3,252	3,621	3,335	1,959	2,942
53	シェーグレン症候群	19,290	–	15	351	950	2,268	3,937	4,268	2,813	4,688
1	特発性大腿骨頭壊死症	19,256	–	21	312	1,141	3,140	4,738	4,483	2,337	3,084
	特発性間質性肺炎	18,399	–	3	15	32	203	843	3,529	4,284	9,490
	特発性拡張型心筋症	18,234	1	14	247	671	2,032	3,861	4,580	2,769	4,059
	原発性胆汁性胆管炎	16,625	–	–	16	155	922	2,646	4,549	3,197	5,140
3	特発性[illegible]血小板減少性紫斑病	16,599	2	70	555	859	1,347	2,033	2,793	2,337	
84		15,627	1	13	78	456	1,469	2,551	4,140		
		15,157	–	56	782	1,731	3,058	3,501	2,51		
		13,544	13	298	876	1,564	3,119	3,749	2,0		
		13,354	–	98	1,260	1,860	3,000	3,149	2		

62 rare diseases in scope

Rare disease sample N = 160

Reference group N = 40

Aiming to launch at Expo 2025

NOTE: we are currently in the stage of finalising the survey and preparing for fielding

CRA Charles River Associates