ECONOMIST IMPACT

Leaving no one behind

Optimising rare disease management in Europe

Background

According to the European Union (EU), rare disease (RD) is a medical condition that affects fewer than or equal to 1 in 2000 persons

≤1 in 2000

There are over 7,000 RDs affecting over 300 million people globally, of which 30 million span across EU alone.[1]

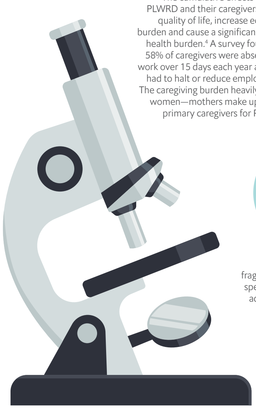
7,000

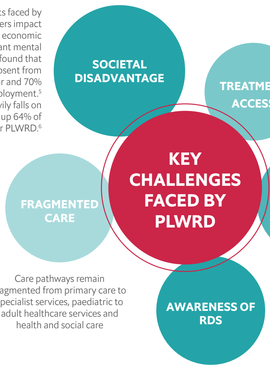
The yearly cost for 15.5 million people impacted by 379 RDs amounted to US$ 1trn in the US alone; the cost in Europe is yet to be determined as studies are lacking.[2]

US$1^{trn}/year

The key challenges faced by persons living with a rare disease (PLWRD)[3]

The cumulative effects faced by PLWRD and their caregivers impact quality of life, increase economic burden and cause a significant mental health burden.[4] A survey found that 58% of caregivers were absent from work over 15 days each year and 70% had to halt or reduce employment.[5] The caregiving burden heavily falls on women—mothers make up 64% of primary caregivers for PLWRD.[6]





Drug treatments (orphan medicinal products) are only available for 5% of the 7,000 RDs (representing 80% of the global RD population) but equitable access remains a key challenge.

PLWRD in Europe wait 5-30 years between early symptoms and an accurate diagnosis.

Care pathways remain fragmented from primary care to specialist services, paediatric to adult healthcare services and health and social care

Healthcare professionals such as primary care physicians report lower awareness of RDs. This ultimately lengthens the patients' diagnostic odyssey and access to treatment.

In December 2021 the UN General Assembly adopted the first-ever resolution on RDs.[7] It calls for the end of discrimination of PLWRD and to address the disparities they face in accessing healthcare.

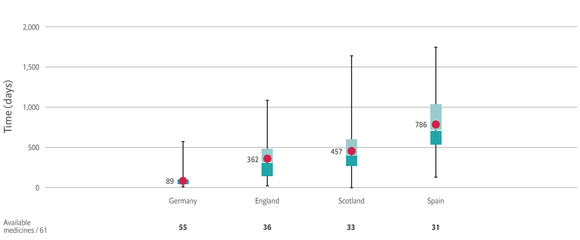
The state of RDs in Europe

Despite the similar number of PLWRD in Germany, Spain and the UK, **disparities exist between countries** in time to diagnosis, availability of treatments, and waiting times from market authorisation to the date of availability to patients.

	GERMANY	SPAIN	UK
Number of PLWRD	4M	3M	3.5M
Average time to diagnosis	6 years	6 years	5 years
Number of RDs in newborn screening (NBS)	19	7	9
Current availability of orphan drugs (out of 61 products authorised in the EU from 2018-2021)	90%	51%	59% ENGLAND, 54% SCOTLAND

Source: References 8-16

The **waiting time for patients to access orphan drugs varies** greatly within and between European countries[16]



STRENGTHS OF RD MANAGEMENT IN EUROPE

GERMANY

provides automatic reimbursement for orphan drugs. On average, patients in Germany wait 3 months to access a new drug treatment from authorisation.[17]

SPAIN

Undiagnosed Rare Disease Programme (SpainUDP) seeks to provide diagnosis for unknown RDs through whole genome sequencing (WGS). The programme has reviewed 335 applications, of which 83 received an accurate diagnosis.[18]

UK

100,000 Genomes Project led over 20,000 participants receiving a new genetic diagnosis, of which 14% found variations in regions of the genome that would be missed by other methods.[19]

Looking ahead towards an equitable future

HARMONISING NEWBORN SCREENING

Over 70% of RDs manifest in childhood therefore early diagnosis is essential for early intervention.[20] Increasing the coverage of RDs screened for via NBS should be a starting point to getting a faster diagnosis.

Germany, Spain and the UK screen for less RDs via NBS, compared to other European countries (Italy screens for 48 RDs).[14]

BETTER DATA COLLECTION AND EVIDENCE GENERATION

The challenge with evidence generation lies with low trial recruitment levels.

Developing central RD registries, expanding international reference and research networks will boost RD awareness, quality of care and access to treatments.

INTEGRATING RD MANAGEMENT INTO EXISTING HEALTHCARE STRUCTURES

Building separate provisions and pathways for RDs may not be financially and economically viable.

Therefore, streamlining RD care pathways that encompass the transition from paediatric to adult services, integrate health and social care, and primary care to specialist services could improve the quality of care.

ENSURING EQUITABLE ACCESS TO TREATMENT

Current HTA processes exacerbate access inequities within and between countries.[17] Having flexible and transparent processes will ensure fair access to promising treatments, regardless of geography.

BIGGER PATIENT VOICE IN DECISION-MAKING

New EU HTA regulation comes into effect in 2028 for RDs, mandating joint clinical assessment which requires patient engagement in scientific consultation and assessment.

Skilling patients in their new role is essential for participation in HTA decision-making; those from marginalised groups must be included in the conversation.[21]

UNRAVELLING THE COST OF INACTION

Quantifying the impact of health systems inefficiencies, such as duplication of appointments, delays in accessing treatment and consequential secondary complications, would provide a comprehensive picture of the cost of inaction towards ensuring true UHC.

To understand the true extent of the RD burden, better research into the cumulative socioeconomic burden of RDs is required.

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