



Rare Diseases
Ireland

Access to Services for Rare Diseases

Annual Business Day
The Pharmaceutical Managers' Institute

4th April 2019
Vicky McGrath, CEO

Rare Diseases - Background

A rare disease is defined in Europe as affecting
fewer than 1 in 2,000 citizens

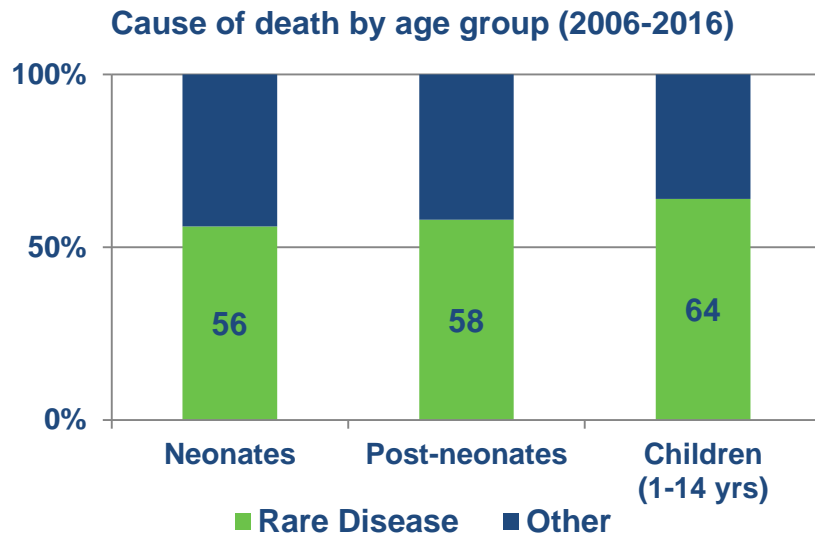
- Over **6,000** distinct rare diseases described
- **25-30 million** people living with a rare disease in Europe
- **70-80%** of rare diseases are genetic in origin
- Onset in **children** for 50% of rare diseases
- Some main groups: metabolic, neuro-muscular, autoimmune, chromosomal anomalies, bleeding disorders, cardiovascular, respiratory, skin diseases, rare cancers, eye diseases, bone disorders, epilepsies, neurological...

Rare Diseases in Ireland

- Estimated 300,000 people will be diagnosed with a RD in their lifetime

Hot off the press.....

- In Ireland
 - Large numbers affected by RDs: by age 17 min incidence 3.3%
 - RDs are serious: 60-65% of paediatric mortality cases (2006-2016) had a RD



Who are the 300,000?



Rare Diseases Ireland

- **Vision:** better lives and cures for people living with a rare disease
- **Mission:** work across all rare diseases to improve the lives of people in Ireland living with a rare disease

Identification – Treatment – Cure

The challenges we face

Most rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening

- Patients & experts are **few**, geographically **scattered** and **isolated**
- Patients are **undiagnosed**, **misdiagnosed** or wait years for diagnosis
- Reliable **information is scarce**
- **Fragmented research**, data and expertise
- Lack of **treatments** and challenges to **access** adequate care
- High social impact and **marginalisation** within society and healthcare systems designed for common diseases
- Heavy **psychosocial burden**

Rare Diseases Ireland: History

- National alliance of patient organisations - a single voice for those affected by or at risk of developing a rare disease
 - 1988 estd. IDO (Inherited Disorders Organisation)
 - GIDO → GRDO → RDI (2017)
 - establishment of national genetics center
 - watchdog against genetic discrimination
 - development of policy to address access to diagnosis and medical and social care

RDI: Active in Europe

- 1999: EU Regulation
Orphan Medicinal Products
- 2008: EU Commission Communication
Rare Diseases: Europe's challenges
- 2009: EU Council Recommendation
Action in the field of rare diseases
- 2011: EU Directive
Patients' rights in cross-border healthcare

- EU Public Health Policy priority
- EU Research Framework Programme priority

RDI: Bringing Europe home...

- 2008: Launch World Rare Disease Day - Last day of February every year
A Rare Day for Rare Diseases
- 2010-2014: EUROPLAN – towards development of a National Plan
 - 2010/11: EuroPlan Conference, Farmleigh multi-stakeholder event
 - 2011: Established Rare Disease (Towards 2013) Taskforce
MRCG, IPPOSI & GRDO (RDI)
 - 2011: Steering Committee for development National Rare Disease Plan
 - 2012: EuroPlan 2
Outcomes of subgroups, surveys and consultation discussed
 - 2013: National Rare Disease Plan finalised
- 2014: Launch National Rare Disease Plan for Ireland 2014-2018

National Rare Disease Plan

- Timely access to best **evidence-based**, patient and family-centred **screening, diagnosis, treatment & care** through all stages of their lives
 - Needs addressed within all aspects of health system
 - Information and research
 - Prevention, diagnosis and care
 - Access to appropriate drugs and technologies
 - Empowering, protecting & supporting patients and carers

Only tackles health aspects of living with a rare disease

Progress: NRDP

- 2014 - National Clinical Programme for Rare Disease
 - 2017 – European Reference Networks
(pooling of expertise at European level)
- 2015 - National Rare Disease Office
- 2018 - Rare Disease Technology Review Committee

A lot done, a lot more to do....

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Social impact of rare diseases

Juggling care and daily life:

The balancing act of the rare disease community

- Europe wide survey, conducted by EURORDIS-Rare Diseases Europe
- Over 3,000 patients and carers
- 802 diseases, 42 countries



A EURORDIS INITIATIVE

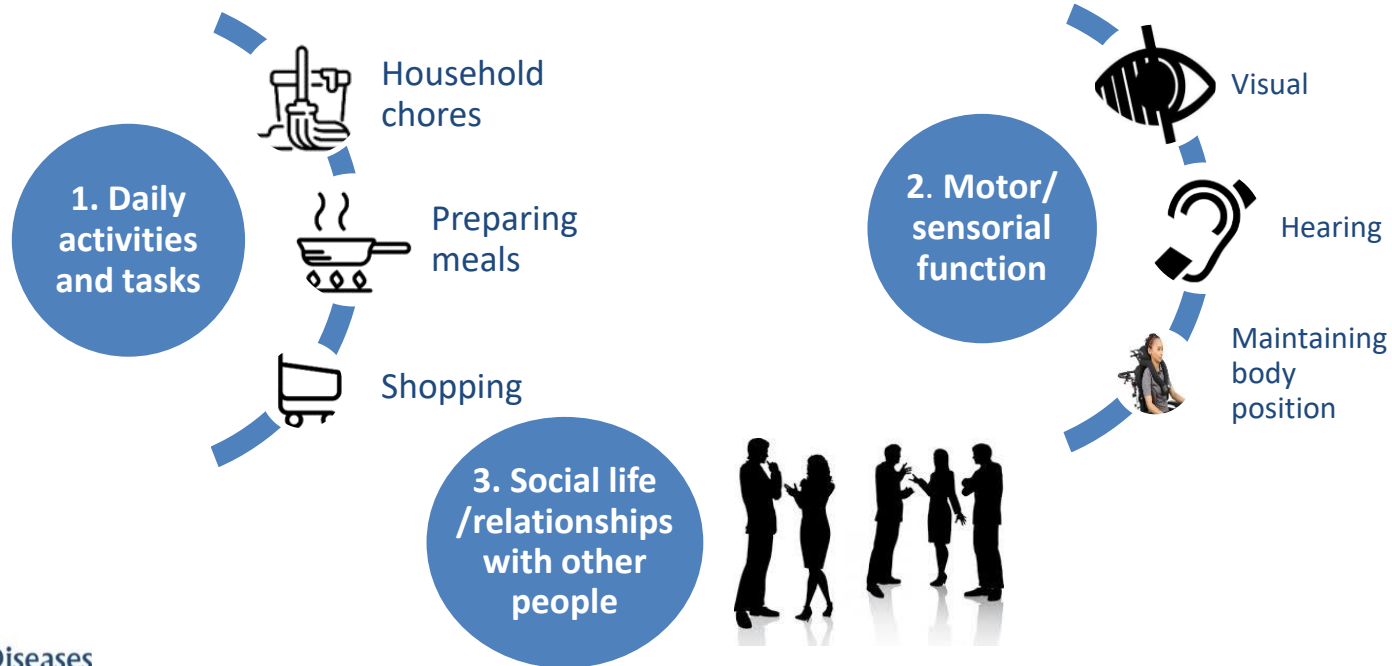


Confirms anecdotal stories

- Serious impact on everyday life
- Significant time and care burden
- Work-life balance: absence from work, hampered professional life, economic burden
- Mental health impact of patients and carers

Serious impact on everyday life

>70% of people living with a rare disease have difficulties



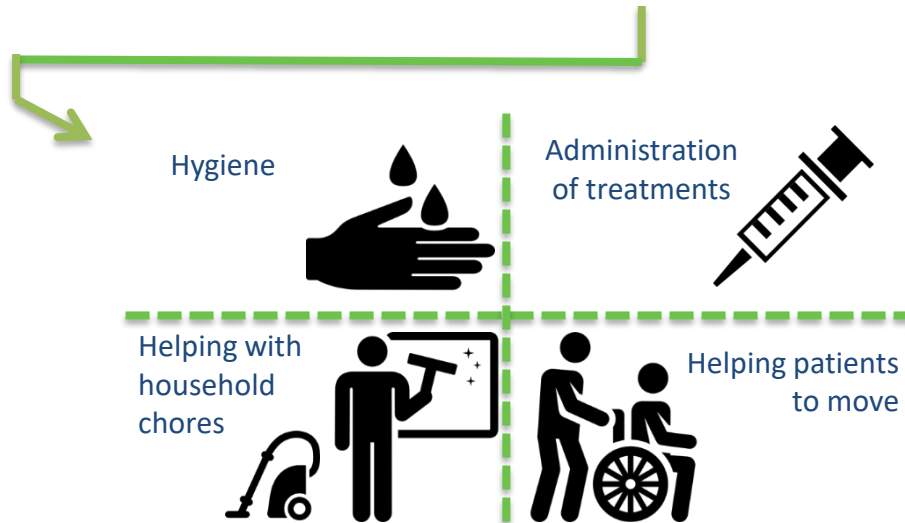
Serious impact on everyday life



8 in 10 people living with a rare disease face **difficulties** in **more than one** aspect of their everyday life

Significant time and care burden

42% of patients & carers spend more than **2h/day** on illness-related tasks



Significant time and care burden

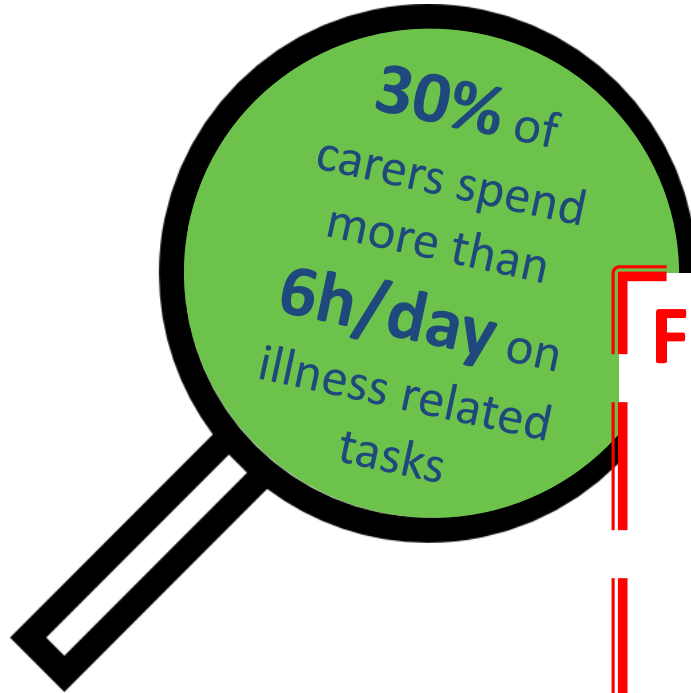
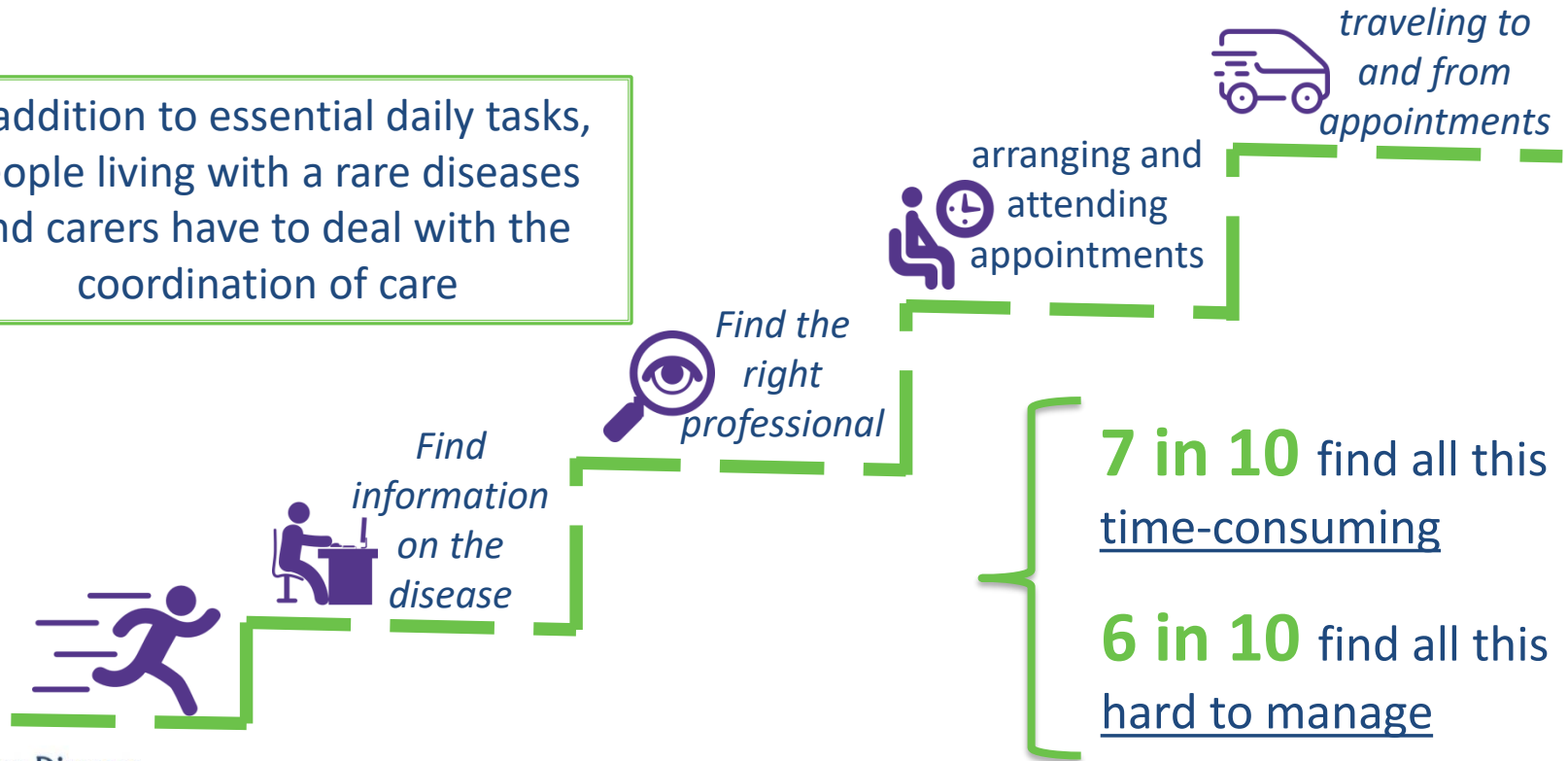


Figure grows to 47% for carers attending severely affected individuals

Organising care is time-consuming

In addition to essential daily tasks, people living with a rare disease and carers have to deal with the coordination of care



Burden falls heavily on women

64% of rare disease patients carers are **mothers**

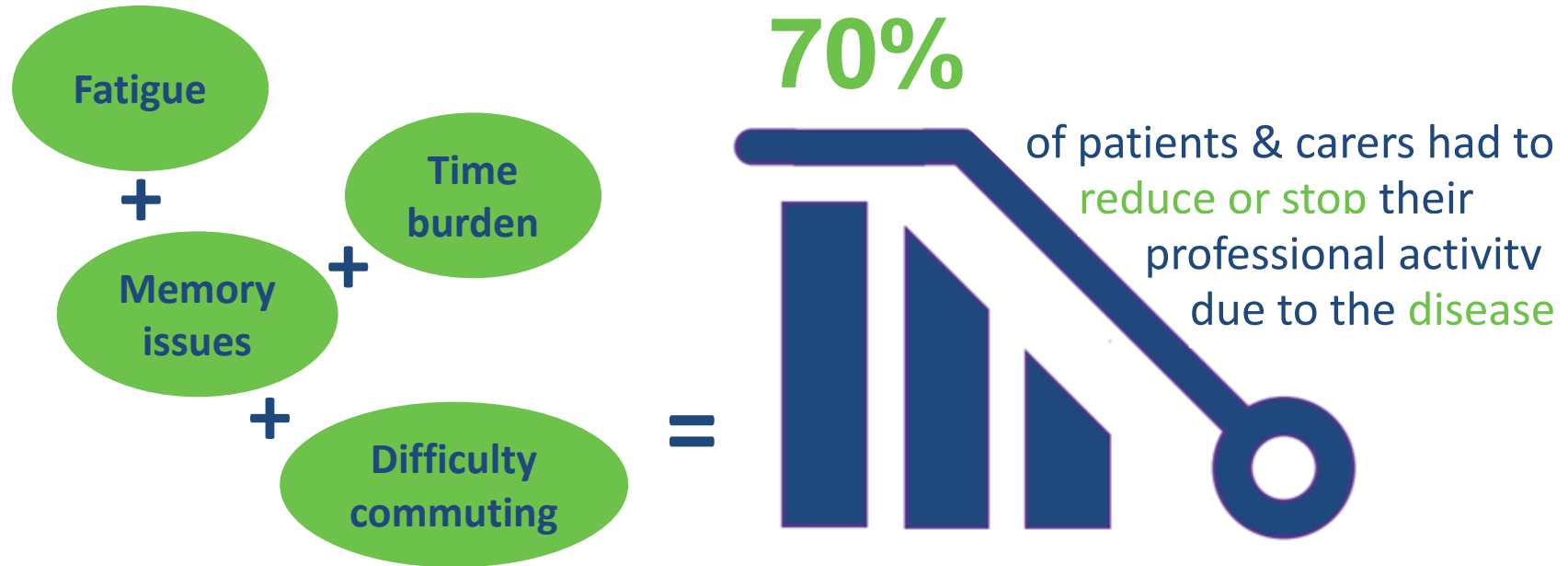


25% of rare disease patients carers are **spouses** (both genders)



The role of the primary carer for people living with a rare disease is primarily **assumed by women**

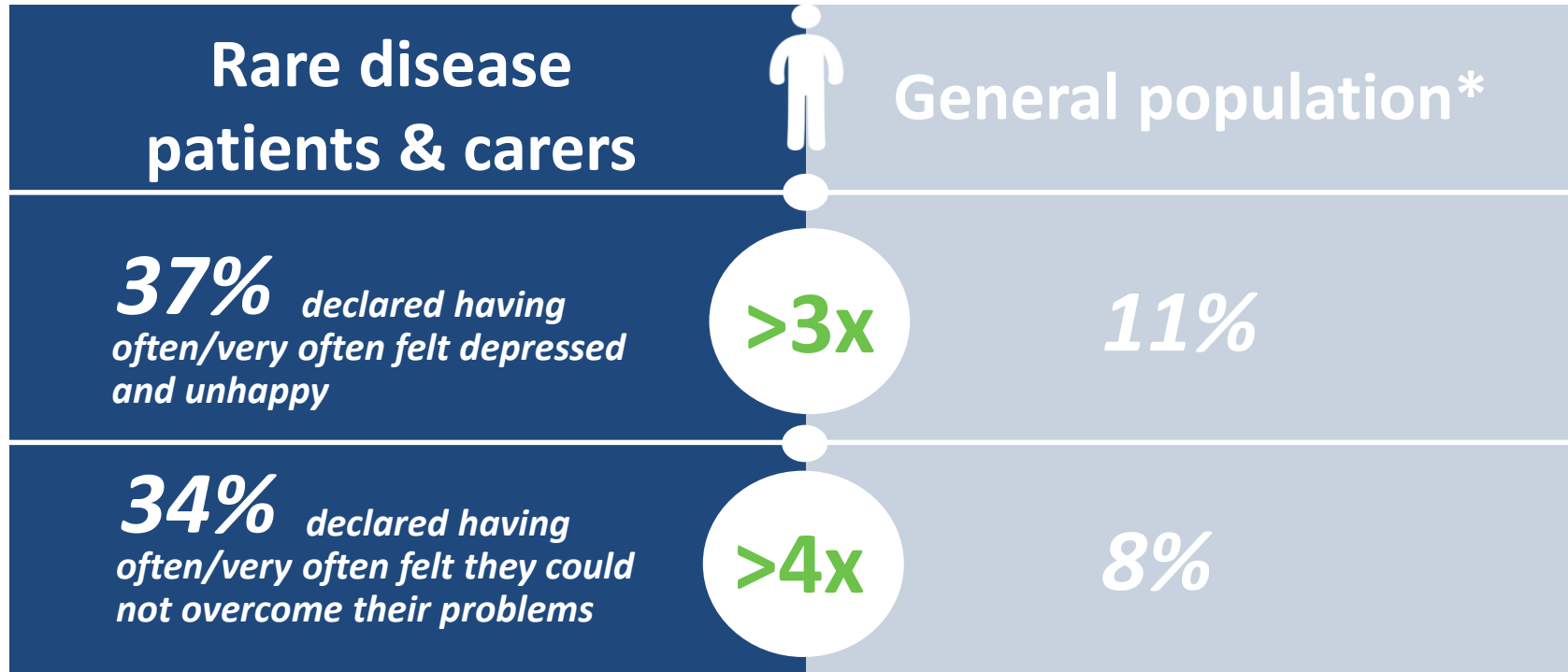
Significant professional life impact



Lack of flexibility in the workplace

- People living with a rare disease often need to stop working during most challenging times
 - **58%** absent from work over **15 days/year**
 - **21%** absent from work over **90 days/year**
- The possibility of asking for special leave is a high unmet need for rare disease patients
 - **41%** asked but received a **negative** response

Mental health of patients & carers



What next for Irish RD patients?

Identification – Treatment – Cure

- Genetic Services
- European Reference Networks
- Access to Medicines



Rethink our approach to care – SLÁINTECARE?

Round Table of Companies

Your voice matters....

- RDI-RTC is a formal & transparent relationship between RDI and industry
 - Semi-annual meetings for RTC members
 - Code of conduct
 - Membership fee

***The patient is at the centre of all of our efforts.
Nothing about us without us!***



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Ireland

THANK YOU!

Vicky McGrath, CEO
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