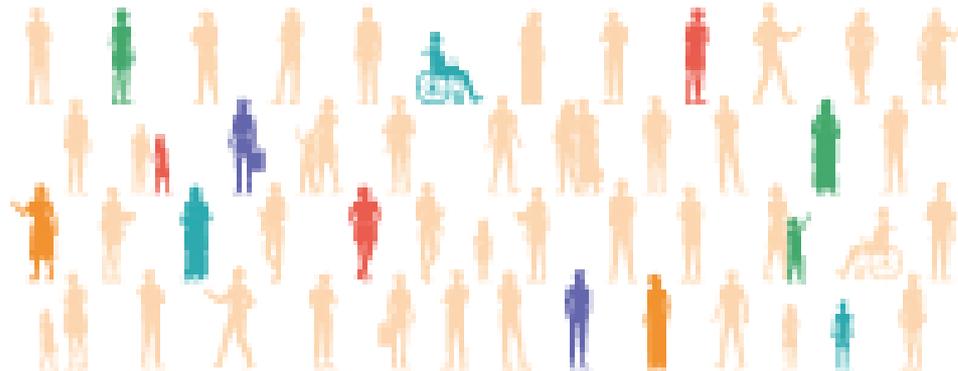


# Rare Diseases Task Force: aims and updates

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## Rare disease

- Several definitions
- Definition EU: any rare disease that affects fewer than 5 people per 10,000 in the EU
- >10 000 rare diseases recognized and



## Epidemiology of rare disease

- Rare diseases affect 3.5% - 5.9% of the worldwide population
- Approx. 30 million people in Europe live with rare disease
- Most are chronic, often progressive and/or degenerative, and life-threatening
- High burden individuals living with rare disease and family
- And on society!



## Epidemiology of rare disease

- Impact of rare diseases is underreported and not comprehensively addressed
- Accurate estimates of rare disease hampered:
  - Medical expertise is often low
  - Difficulty getting diagnosis
  - Health care often inadequate
  - No ICD codes available
  - Etc.

## Burden EU - Rare Diseases Task Force

### Aims:

- Perform research together on the population health burden of rare diseases;
- Ultimate goal to make rare diseases more visible for policy makers, researchers and the general public.
- Founded in spring 2022
- Currently 8 members who meet every 4-6 weeks



## Activities of the task force

- Work on and discuss ongoing projects.
- Ideas for collaboration, joint activities and publications.

### Current projects:

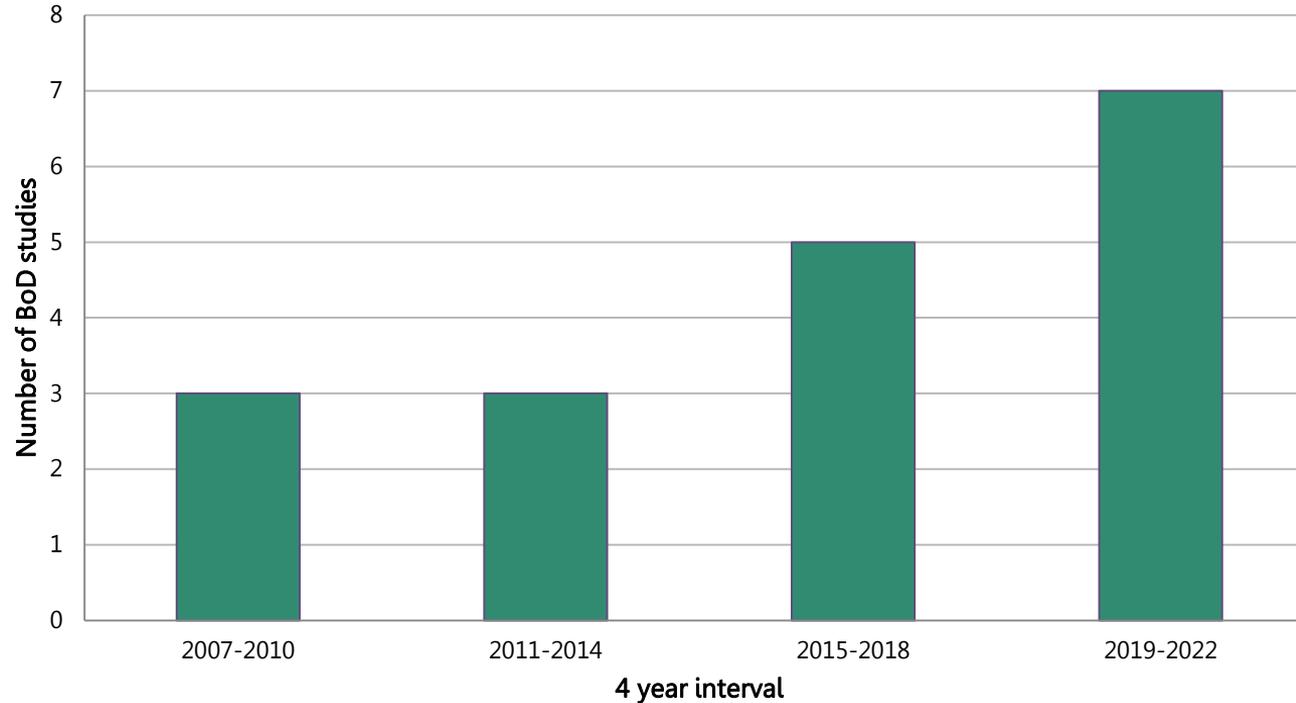
- Systematic literature review of burden of rare disease studies.
- Devising a plan to map burden of disease indicators in rare disease registries across Europe.
- Work on other publications together.

## Systematic literature review

- SLR of studies that assessed the burden of one or more rare diseases (1990-2022).
- Focus on methods and data input sources that were used for the calculations.
- In total >400 records screened
- 17 studies met inclusion criteria (worldwide)
- In comparison, for infectious diseases we found over 90 studies in Europe.



## Number of studies over time



## Rare diseases and GBD study

- Global Burden of Disease study reports DALYs separately for small number of rare diseases (of the >rare 10,000 diseases)
- E.g. Down syndrome, Turner syndrome, multiple sclerosis

THE LANCET

Volume 393 Number 10156 Pages 1170-1200 October 12-18, 2019 www.thelancet.com

The Global Burden of Disease Study 2019



## The global burden of rare diseases

- However, the Global Burden of Disease study lumps many rare diseases together in a single category.
- E.g. “other neurological disease” or “other musculoskeletal disorders”.
- This hampers insight into the burden of rare diseases, also in comparison to other conditions.
- Lack of ICD codes does not help.....

## In summary

- Rare diseases are not rare.
- Burden of disease studies on rare diseases are rare.
- This contributes to the invisibility of rare diseases.
- The Burden EU Rare Diseases Task Force aims to make rare diseases and their impact more visible.



# Questions?

