

Rare Diseases Task Force: aims and updates

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What are rare diseases

- Definition EU: any rare disease that affects fewer than 5 people per 10,000 in the EU
- >10 000 rare diseases recognized
- Rare diseases affect 3.5% - 5.9% of the worldwide population
- Most are chronic, often progressive and/or degenerative, and life-threatening



Rare Diseases Task Force

- Founded in spring 2022
- Currently 16 members who meet every 6-8 weeks

Aims of task force:

- 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.

Activities of the task force

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

Projects:

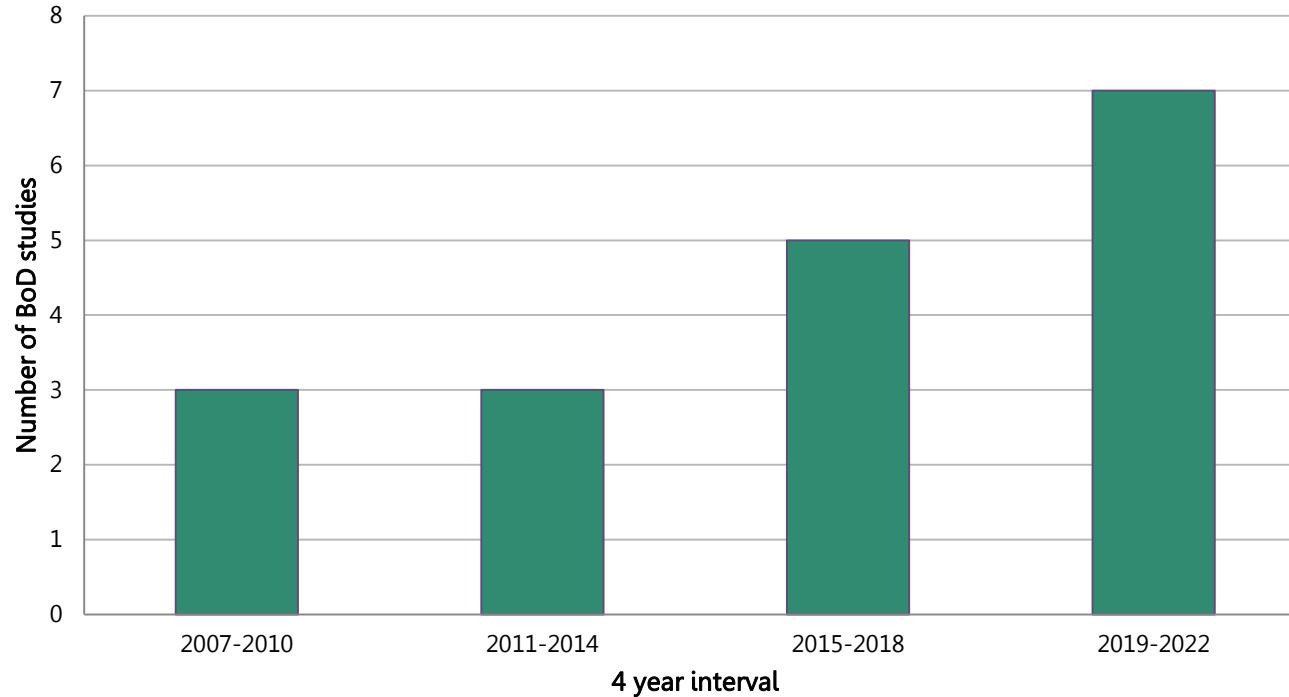
- Systematic literature review of burden of rare disease studies.
- Map burden of disease indicators in rare disease registries across Europe. (presentation Diane Grad)
- Determine the burden of SMA

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



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.
- If you want to join the Task Force, email:
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Questions?

