

BURDEN OF DISEASE: WHAT IS IT AND WHY DOES IT MATTER FOR RARE DISEASES?

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Lifestyle and Chronic Diseases

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Key questions public health

- What are the most relevant diseases in a country?
- Which risk factors are the strongest contributors to disease and death?
- How is the impact of different diseases evolving over time?
- How does it compare between and within countries?

Which disease is most important?



low back pain



*coronary
heart disease*



common cold

Burden of disease

How to define disease “impact” or “importance”?



Disease have an impact on multiple aspects

- Burden of disease = quantification of any of these aspects
- This requires specific metrics!

Burden of disease

Which disease is most important?

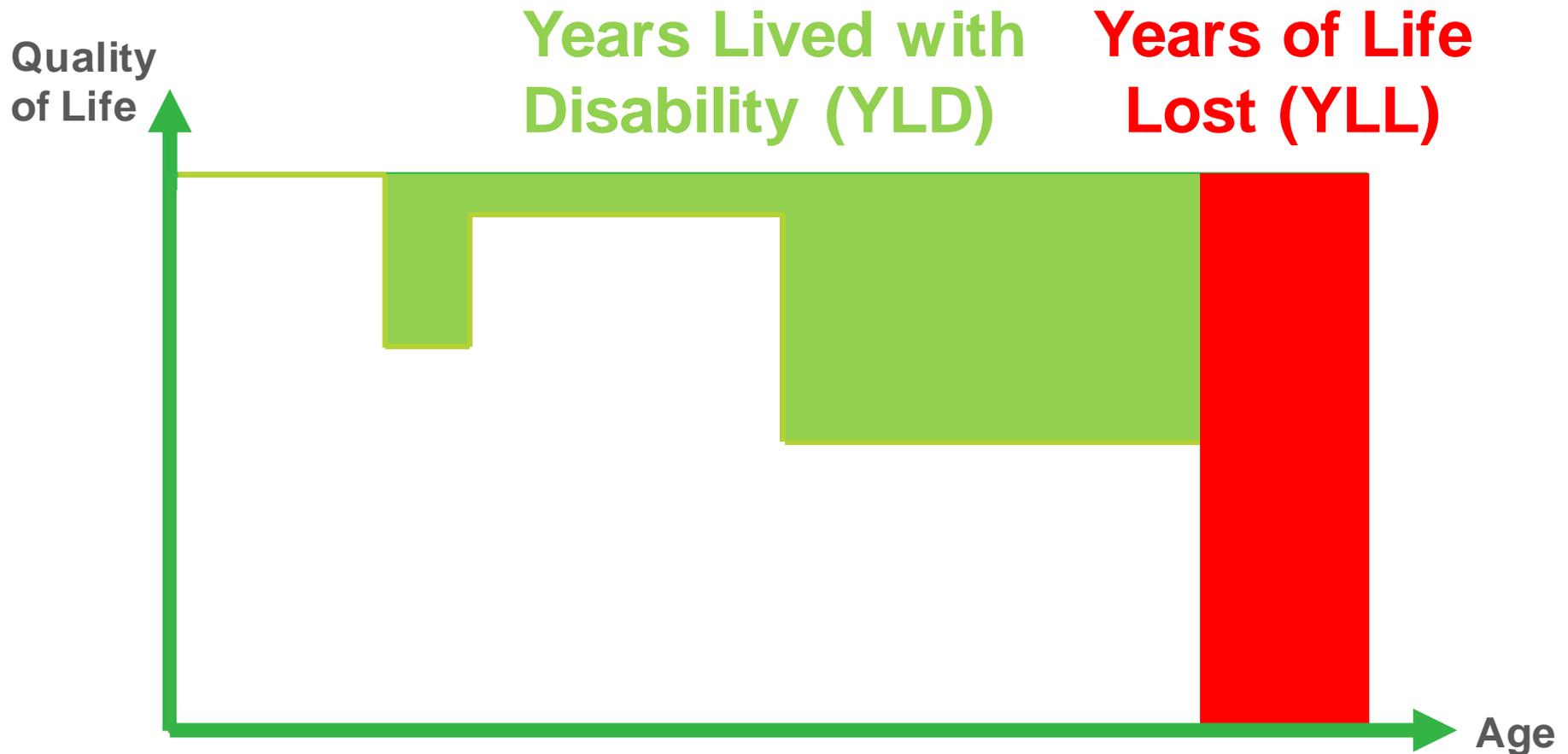


Number of cases, **number** of deaths

- ⇔ **Severity** of case: duration, reduction quality of life
- ⇔ **Severity** of death: residual life expectancy

→ **Summary Measures of Population Health**

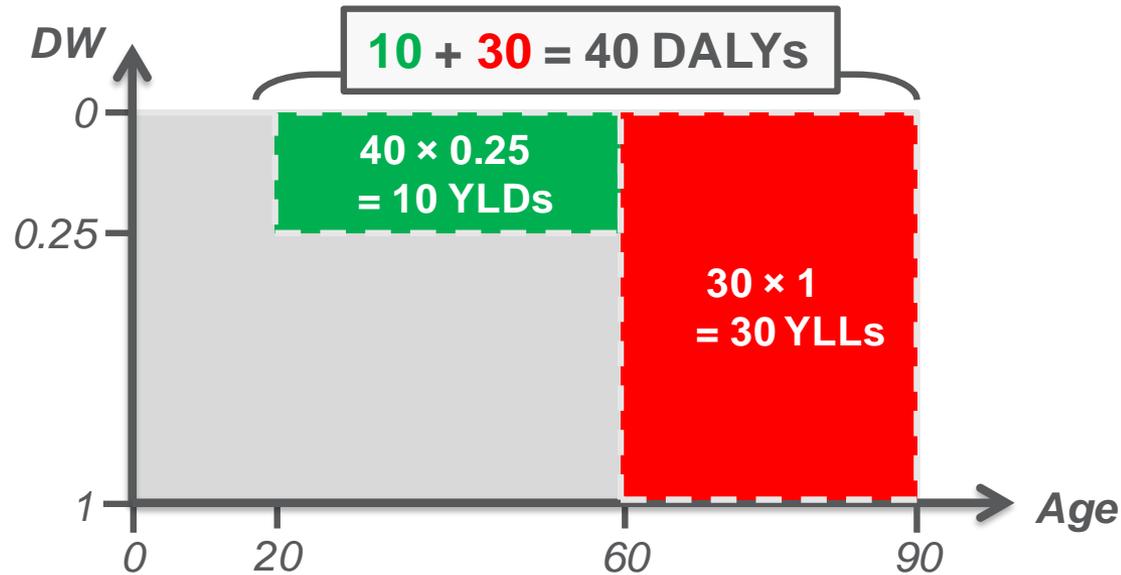
Disability-Adjusted Life Years



Disability-Adjusted Life Years

- **1 DALY = 1 healthy life year lost**
- Summary measure of population health
 - Morbidity + mortality
 - Disease occurrence + disease severity
- **DALY = YLD + YLL**
 - **YLD** = Years Lived with Disability
= Number of incident cases × Duration × Disability Weight
 - **YLL** = Standard Expected Years of Life Lost
= Number of deaths × Residual Life Expectancy

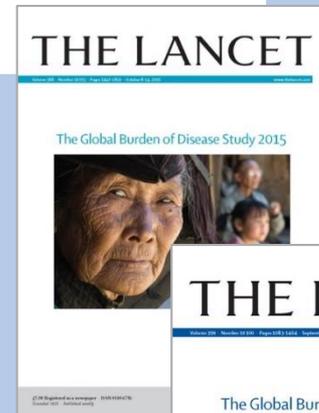
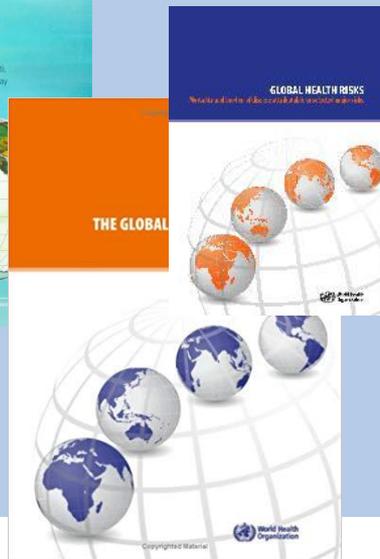
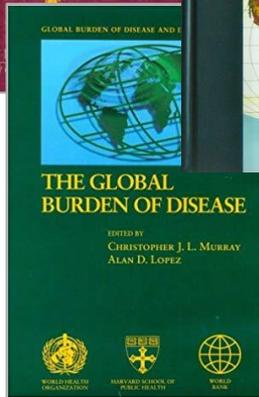
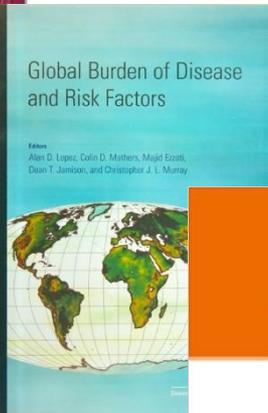
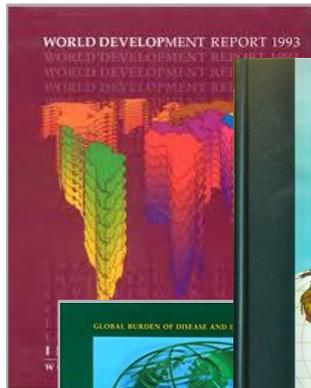
Disability-Adjusted Life Years



$$\text{DALY} = \text{YLD} + \text{YLL}$$

- YLD = Years Lived with Disability = $N \times D \times DW$
- YLL = Years of Life Lost = $M \times RLE$

Global Burden of Disease



THE LANCET

Volume 390, Number 10207, Pages 1151-1201, October 11, 2017 www.thelancet.com

The Global Burden of Disease Study 2019



The Global Burden of



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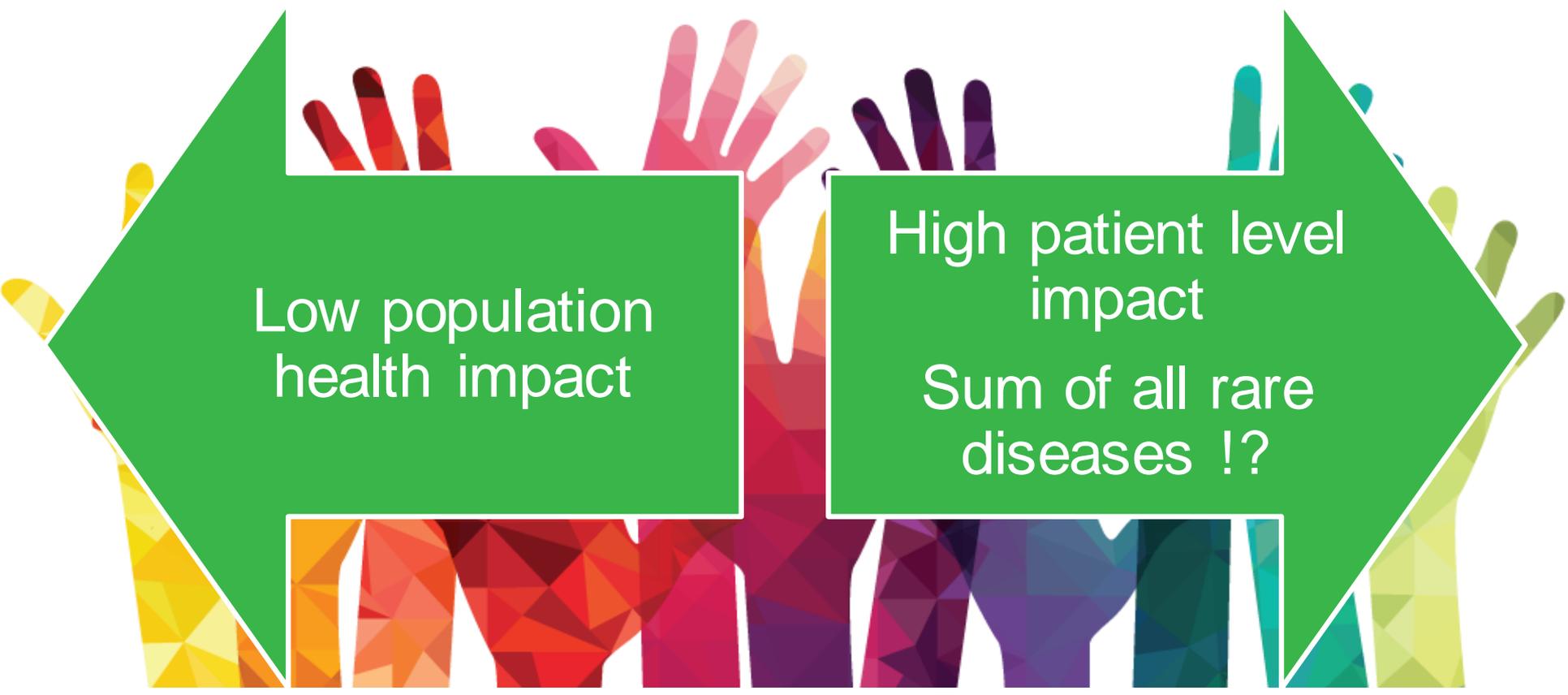


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of disease
network



www.burden-eu.net

Burden of rare diseases ?



Low population
health impact

High patient level
impact

Sum of all rare
diseases !?

GBD and rare diseases

Hemoglobinopathies and hemolytic anemias	D55-D61.9
Thalassemias	D56-D56.3, D56.5-D56.9, D57.4-D57.419
Thalassemias trait	
Sickle cell disorders	D57-D57.3, D57.8-D57.819
Sickle cell trait	
G6PD deficiency	D55-D55.2
G6PD trait	
Other hemolytic anemias	

Case definition

Motor neuron diseases (MND) are a set of chronic, degenerative, and progressive neurological conditions typified by the destruction of motor neurons and the subsequent deterioration of voluntary muscle activity. The most common MND is amyotrophic lateral sclerosis. The ICD-10 code corresponding to motor neuron diseases is G12. Our gold standard diagnostic criteria are the El Escorial Criteria, with other similar criteria (eg, the original set from World Federation of Neurology) if necessary.

Other musculoskeletal disorders	I27.1, L93-L93.2, M00-M03.6, M07-M09.8, M11-M15.9, M18-M25.9, M30-M32.9, M34-M36.8, M40-M43.9, M45-M46.99, M61-M63.89, M65-M68.8, M70-M72.4, M72.8-M73, M75-M77.9, M79-M79.676, M79.8-M87.09, M87.2-M89.59, M89.7-M95.9, M99, M99.05-M99.09, M99.15-M99.19, M99.25-M99.29, M99.35-M99.39, M99.45-M99.49, M99.55-M99.59, M99.65-M99.69, M99.75-M99.79, M99.85-M99.9
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National burden of rare diseases

Café et al. *Orphanet Journal of Rare Diseases* (2019) 14:211
<https://doi.org/10.1186/s13023-019-1175-5>

Orphanet Journal of
Rare Diseases

RESEARCH

Open Access

Haemophilia A: health burden

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Patrícia R. Pinto^{7,8},



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AMYLOID
<https://doi.org/10.1080/13506129.2019.1701429>

ARTICLE

Societal costs and burden of hereditary transthyretin amyloidosis polyneuropathy

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DISEASES

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The health and economic burden of haemophilia in Belgium: a rare, expensive and challenging disease

Séverine Henrard^{1,2*}, Brecht Devleeschauwer^{1,3}, Philippe Beutels⁴, Michael Callens⁵, Frank De Smet^{5,6},
Cedric Hermans² and Niko Speybroeck¹

Burden of rare diseases

Lack of evidence on burden of rare diseases

GBD study: limited and unspecific

National studies: very limited

↔ RD registers offer wealth of information to calculate disease burden for individual patients

EBoDN will encourage and support researchers to initiate rare disease burden assessments

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Health indicators unit

Lifestyle and chronic diseases

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