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BURDEN OF DUCHENNE MUSCULAR DYSTROPHY (DMD) IN BELGIUM

Marjan Cosyns, Eva De Meulemeester, Brecht Devleesschauwer

EJP RD - Measuring burden of rare diseases workshop, 14/09/2021

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DMD: Symptomatology

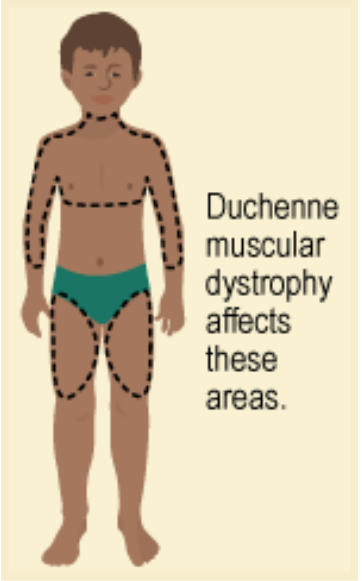
Severe, progressive, muscle-wasting disease

2-3y
- Difficulties climbing stairs
- Waddling gate
- Frequent falls

10-12y
- Wheelchair dependence

~20y
- In need of assisted ventilation

20-40y
- Death from cardiac and/or respiratory failure



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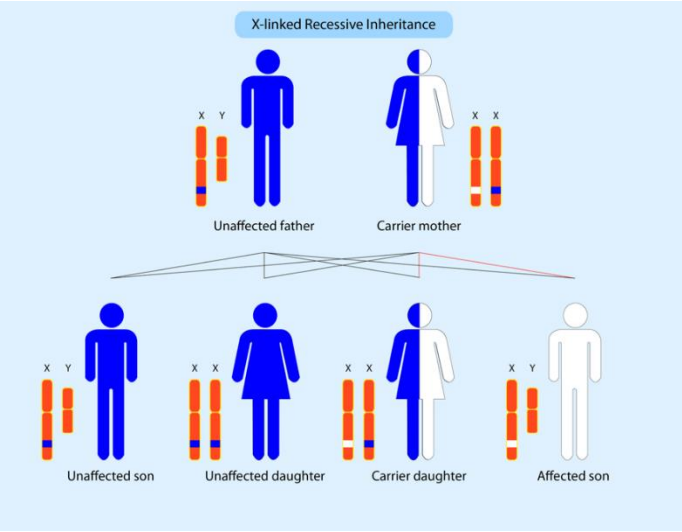
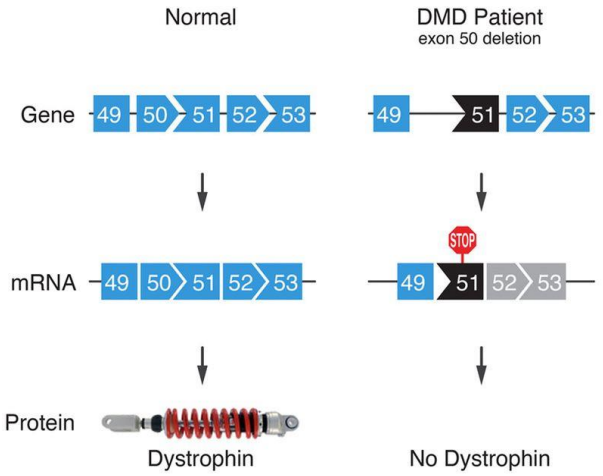
DMD: Genetics

X-linked recessive disorder

- Mutations in the gene encoding dystrophin (*DMD*; 300377)
- Primarily affects boys
- Women are carriers, but can be symptomatic too





DMD: Care in Belgium

Part of the rehabilitation agreement « Neuromuscular diseases (NMD) » (NIHDI, 1999)

- Accreditation of neuromuscular reference centers (NMRC)
  - Expertise
  - Multidisciplinarity
  - Accurate registration
  - Patient-oriented scientific research
- Reimbursement of costs for NMD patients
- Basis of the Belgian Neuromuscular Diseases Registry (BNMDR, 2008)

BNMDR

Annual data collection

7 NMRC

~ 6000 patients/year

62 NMD groups

Demographic data
Diagnosis
Functional status (+ PROM)
+ DMD core dataset
+ SMA core dataset

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TREAT-NMD Neuromuscular Network

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DMD: BNMDR 2019 data

N = 258

- 4% of BNMDR 2019 population
- **~ prevalence: 2.3 / 100,000**

- Mean age = 18y (SD = 9.4y)
- 99% male
- 93% genetically confirmed

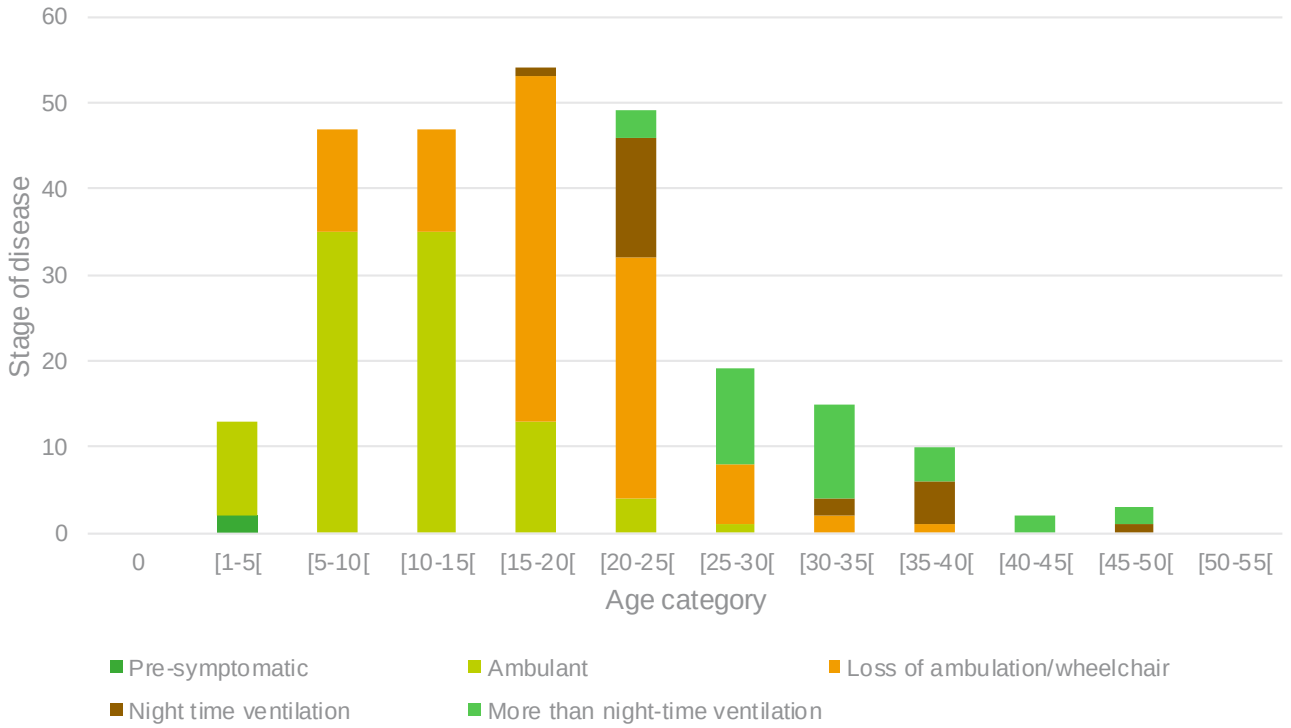


Figure 1. Stage of disease in function of age

Objective of the study

As a rare disease, DMD has a small health impact at population level

However, the health impact at patient level is significant

Need to measure the burden of DMD

- Health care planning
- Emerging gene therapies

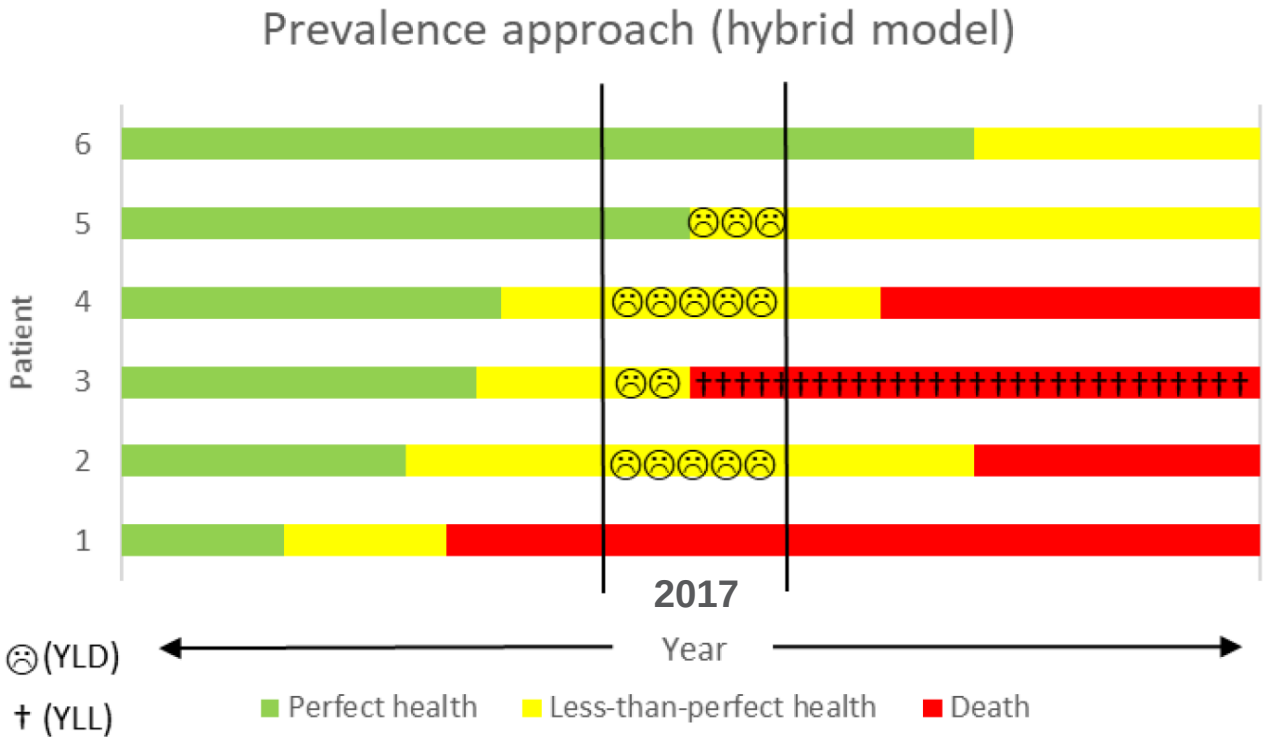
The current study aimed to quantify the burden of DMD in Belgium using the Disability Adjusted Life Year (DALY) metric, based on the BNMDR

Methods: General concepts

DALY = YLL + YLD

- YLL = Number of deaths x life expectancy at the age of death
- YLD = Number of cases x duration till remission or death x disability weight

> BNMDR 2017, 2018, 2019; averaged out



Methods: YLL

YLL = Number of deaths x life expectancy at the age of death

BNMDR mortality data

GBD 2017 study

Methods: YLD

YLD = Number of cases x duration till remission or death x disability weight

- BNMDR data
- Literature
- Scenario analysis

- No specific DW for DMD
- Disease model
  - GBD studies
  - European DW study
  - Literature

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Results: YLL

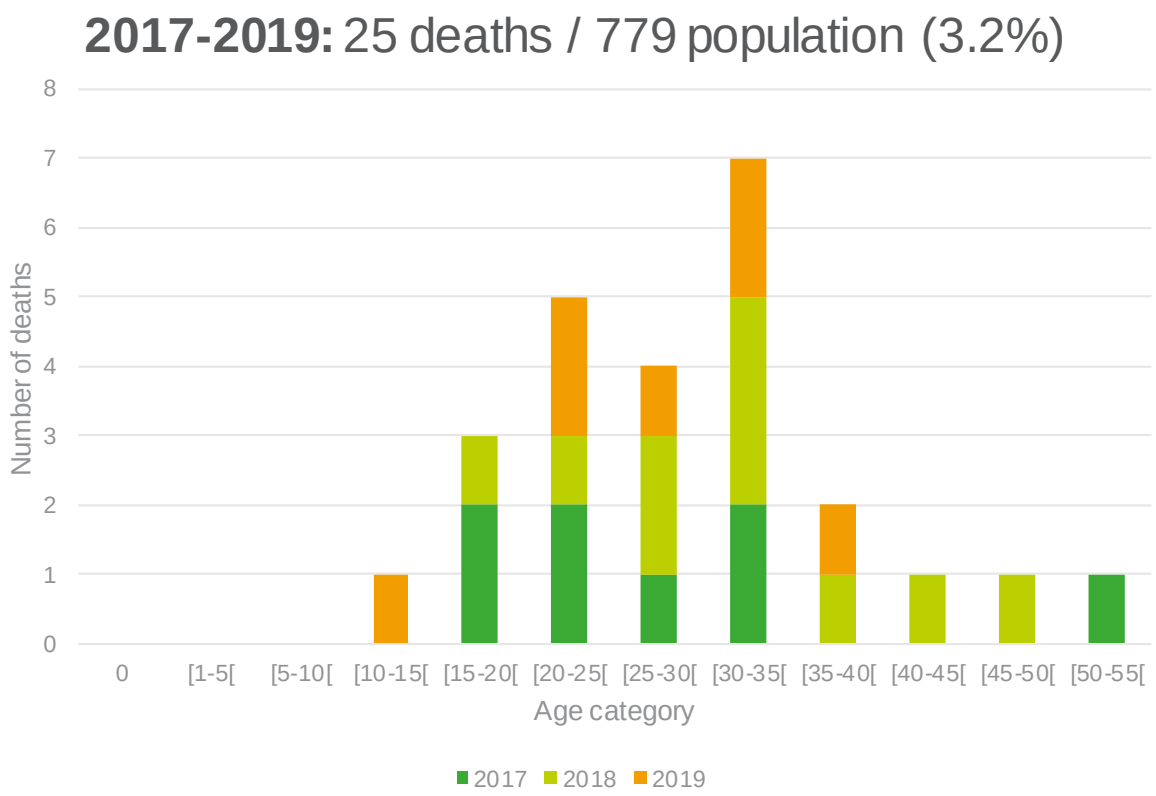

Figure 2. Number of deaths in function of age, per year

$\mathbf{YLL_{2017\text{-}2019}} = \sum_{i=25} YLL\ per\ patient = 1533$

$\mathbf{YLL_{per\ year}} = YLL_{2017\text{-}2019} / 3 = 511$

$\mathbf{YLL_{per\ patient}} = YLL_{2017\text{-}2019} / 779 = 1.97$

$\mathbf{YLL_{per\ death}} = YLL_{2017\text{-}2019} / 25 = 61$

Highest YLL in 30-35y age category

0 YLL before 10y

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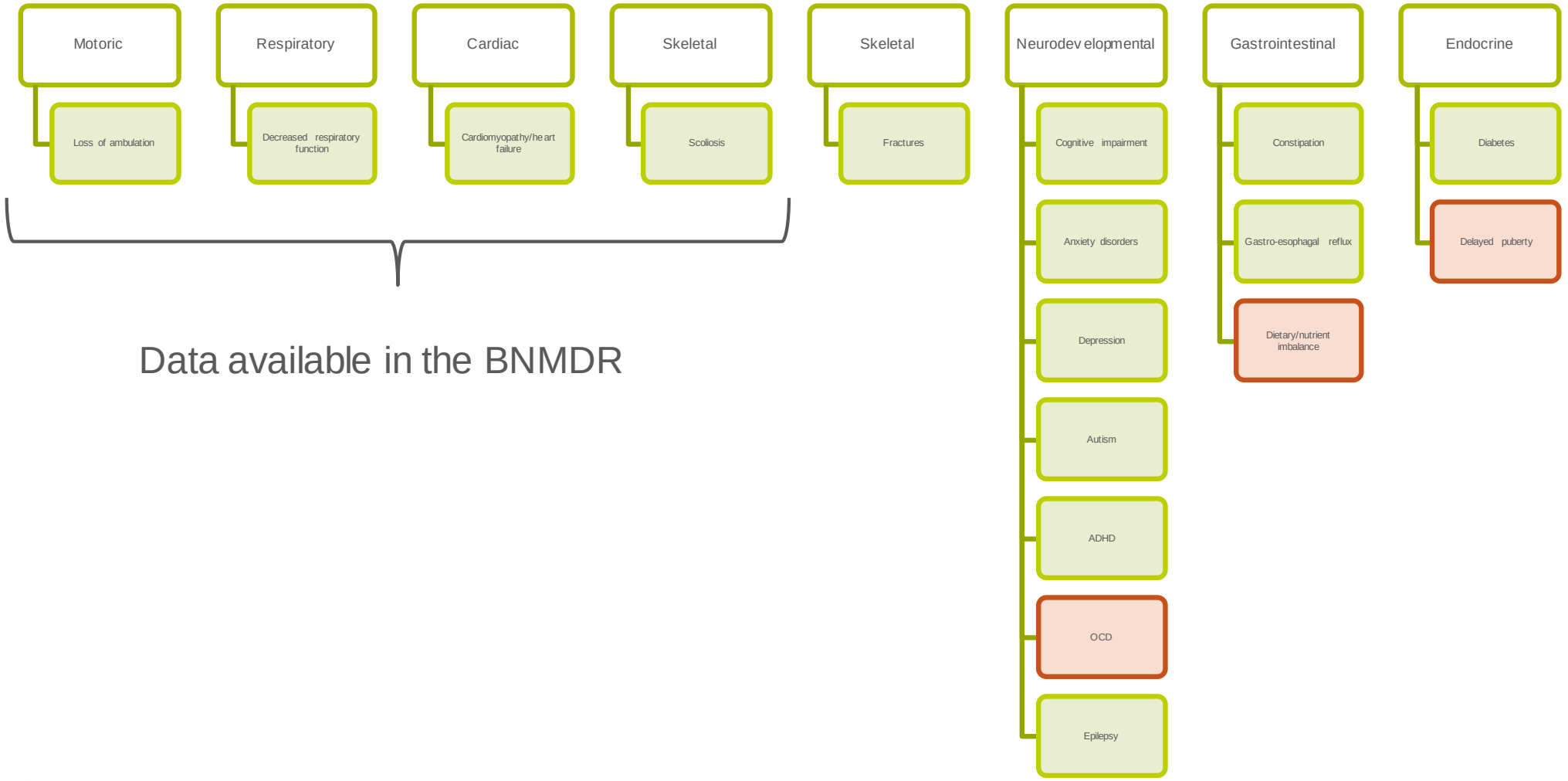
Results: DMD disease model



Data available in the BNMDR

Results: YLD

$\mathbf{YLD_{2017\text{-}2019}} = \sum_{i=779} YLD\ per\ patient = 269$

$\mathbf{YLD_{per\ year}} = YLD_{2017\text{-}2019} / 3 = 90$

$\mathbf{YLD_{per\ patient}} = YLD_{2017\text{-}2019} / 779 = 0.35$

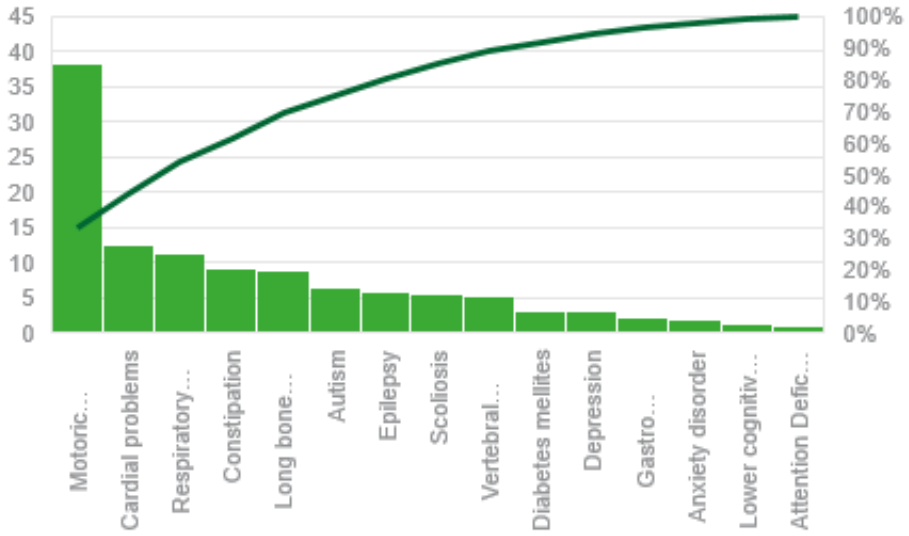


Figure 3. YLD contribution per health state <2017-2019>

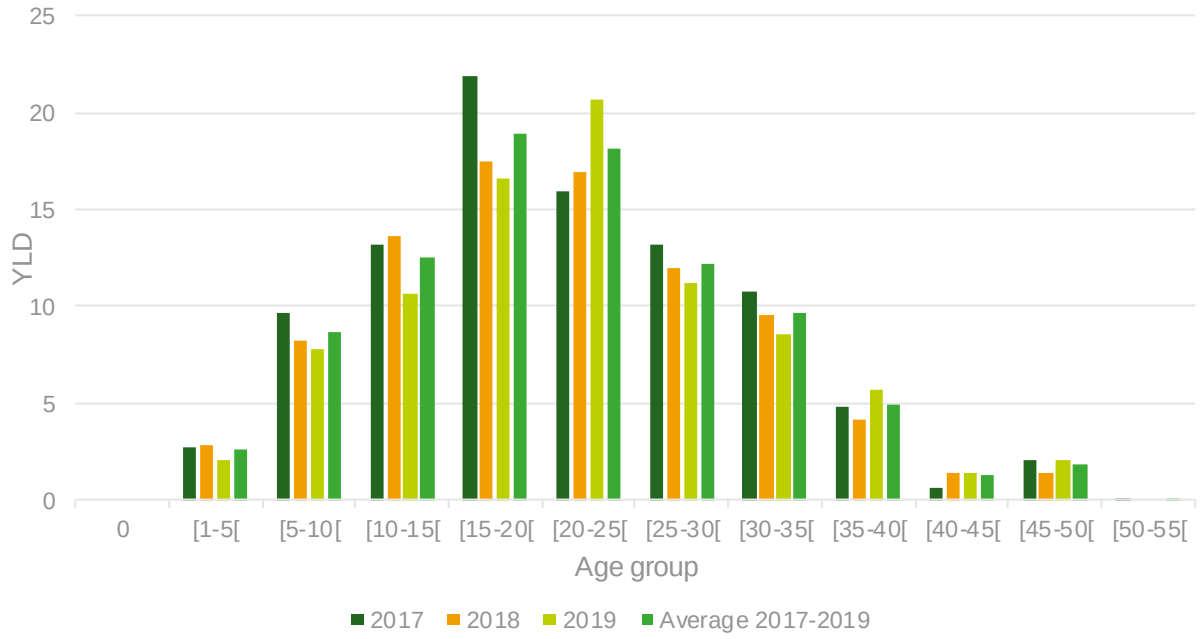


Figure 4. YLD in function of age, per year and averaged out

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Results: DALY

$$DALY_{2017\text{-}2019} = YLL_{2017\text{-}2019} + YLD_{2017\text{-}2019}$$
$$= 1533 + 269$$
$$= 1802$$

$$DALY_{per\ year} = DALY_{2017\text{-}2019} / 3 = 601$$

$$DALY_{per\ patient} = DALY_{2017\text{-}2019} / 779 = 2.31$$

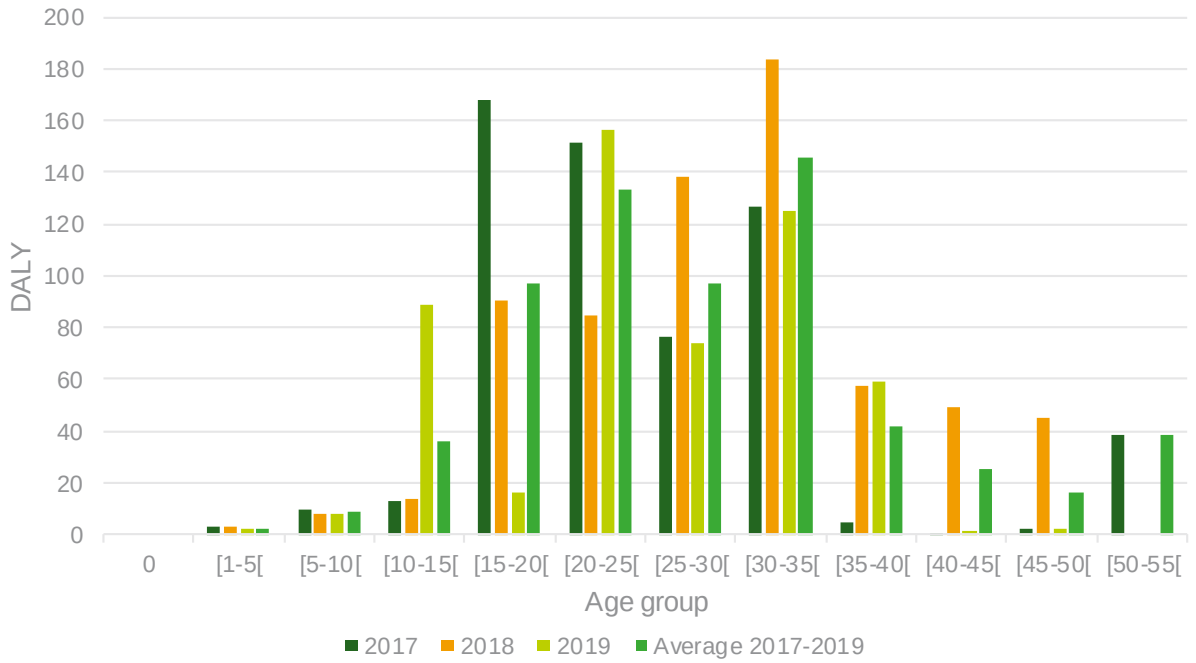

Figure 5. DALY in function of age, per year and averaged out

Results: Scenario analysis

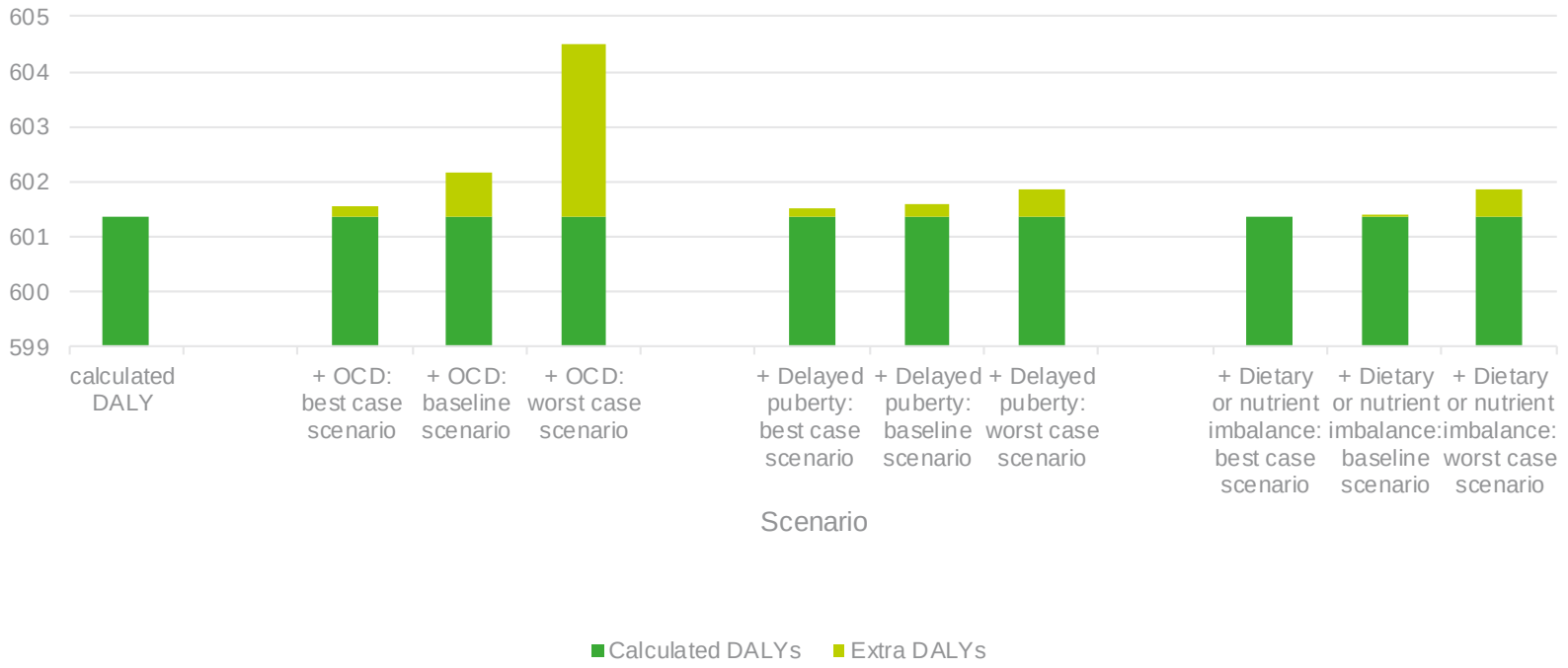

Figure 6. Scenario analysis of the DALY contribution of OCD, delayed puberty and dietary/nutrient imbalance

Discussion

DMD: 2.31 DALY per patient (85% YLL, 15% YLD)

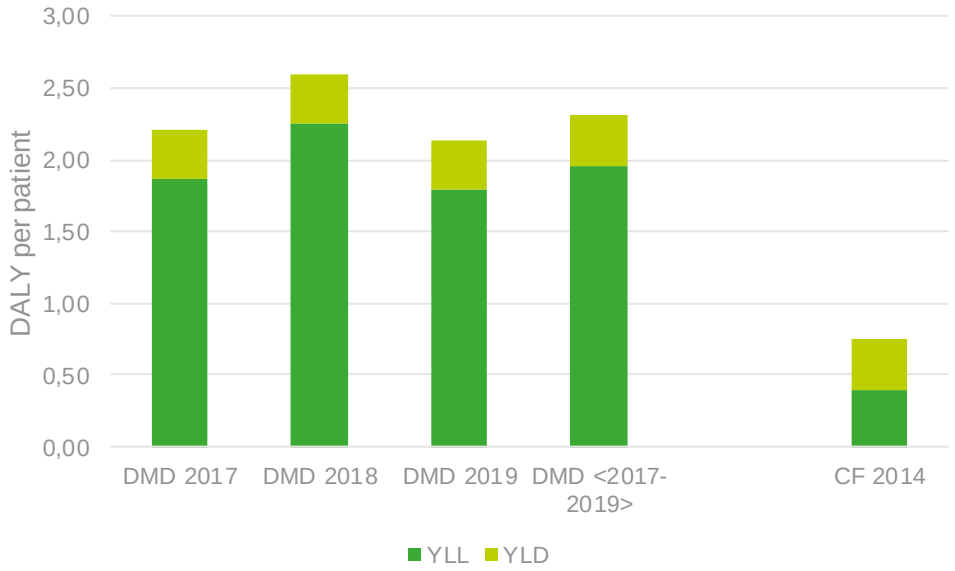

Figure 7. DALY per patient for DMD and CF

CF: 0.75 DALY per patient (53% YLL, 47% YLD)

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Discussion

Strengths:

- First study
- Reliability
- External validity

Future perspectives:

- Dataset modification
- Comparison
- Health care planning

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Contact

Marjan • Cosyns@sciensano.be • +32 2 642 54 15

Sciensano • Rue Juliette Wytsmanstraat 14 • 1050 Brussels • Belgium
T +32 2 642 51 11 • T Press +32 2 642 54 20 • info@sciensano.be • www.sciensano.be

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