

Gap analysis of 4-year data in the Dutch Dystrophinopathy Database

INTRODUCTION

Disease registries are important for the acquisition of real-world data. They can be used for natural history studies, facilitation of trials, and the implementation of therapies in national healthcare systems. It is essential to capture high quality data that reflect consistent and standardized outpatient follow-up according to standards of care.

AIM

To conduct a gap-analysis of standardized clinical data collected in the Dutch Dystrophinopathy Database (DDD)^{1,2} in patients diagnosed with Duchenne muscular dystrophy (DMD).

METHOD

Participants: 254 DMD patients with a DMD diagnosis according to the hospital coding systems of LUMC and Radboudumc between 2018 and 2021.

Categorization:

Loss of ambulation (LoA) defined as the inability to walk 10 meters unaided.

- Assessment performed and recorded in the DDD
- Assessment not performed, noted in the DDD
- Missing data where an entry was expected based on attendance.

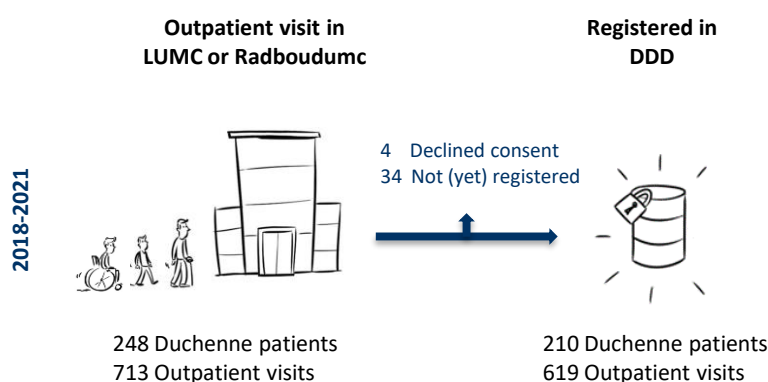


Fig 1. Number of participants and their outpatient visits included in the gap analysis

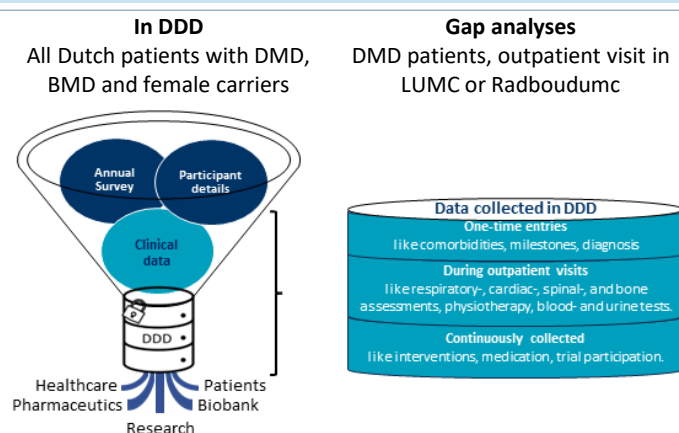
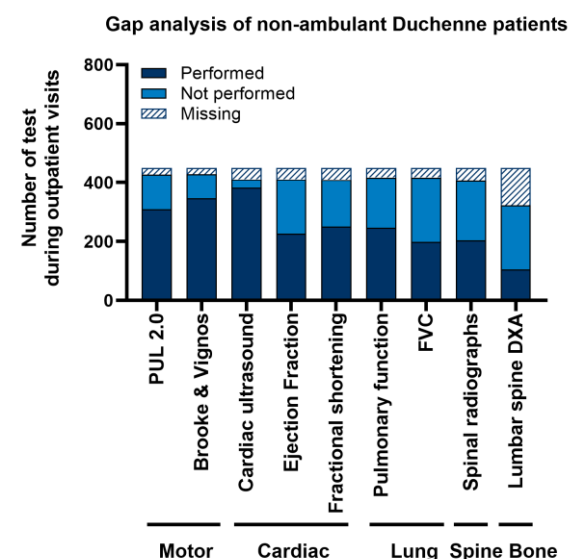
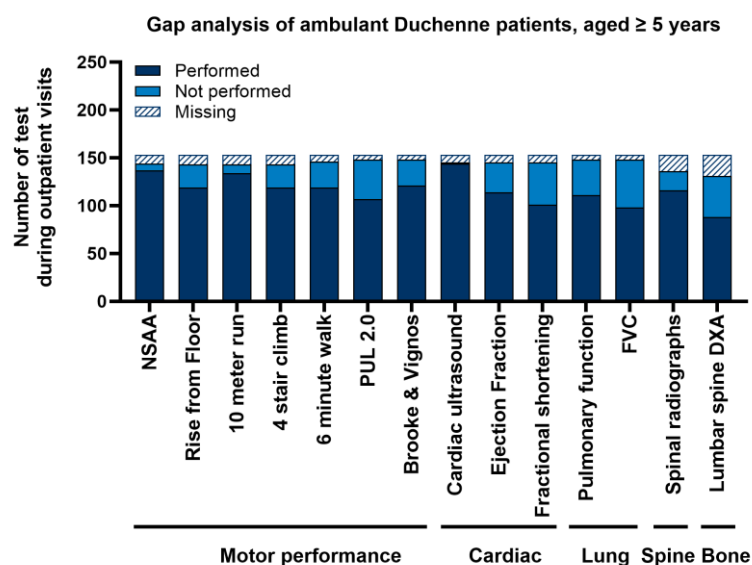


Fig 2. Inclusion in DDD and the section used for the gap analysis. The DDD gathers nationwide self-reported data and clinician-reported data from patients at LUMC and Radboudumc. The gap analysis was conducted using the clinician-reported data.

RESULTS

DDD coverage was high: 85% of DMD patients at Radboudumc and LUMC registered.

- Data on genetics, corticosteroid use, and ambulatory status was 100% captured.
- Missing data reveals an average of 5.4%.
- DXA scans were not performed at Radboudumc
- Performance for PUL 2.0 decreased from 86% before age 18 to 52.3% aged ≥ 18
- Pulmonary function dropped from 72.5% to 54.9% after LoA



RESULTS Ambulant patients

- NSAA performed in 90%, Cardiac ultrasound in 94% and pulmonary function tests in 73% of the visits.
- At LUMC, DXA scans were completed in 83% of the visits

RESULTS non-ambulant patients

- PUL 2.0 performed in 69%, Cardiac ultrasound in 85% and pulmonary function in 55% of the visits.
- At LUMC, DXA scans completed in 79% under 18 and 6% aged ≥ 18 , with no missing data

DISCUSSION

Most gaps were a result of the transitioning from pediatric to adult care and referral patterns within the Dutch healthcare system. For example, patients with an FVC $<60\%$ are seen by home ventilation centers outside DCN. Only minor gaps arose from center to center variability.

CONCLUSION

- The DDD provides consistent coverage of real-world data in the Netherlands especially in the ambulant pediatric population.
- Gap analyses are essential to assess the quality of disease registries and provide insights for quality improvement.

References: (1) van de Velde NM, Krom YD., The DDD: A National Registry with Standardized Patient and Clinician Reported Real-World Data. J Neuromuscul Dis. 2024;11(5):1095-1109.

(2) **Poster 602:** FISMA for high quality and interoperable real-world data on Dystrophinopathies; **Poster 609:** Integration of healthcare and research to optimize treatment and collect real-world data in NMD