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The Dutch Dystrophinopathy Database: facilitation of trial readiness and effective use of patient data

Introduction

The **Dutch Dystrophinopathy Database (DDD)** is a register for all Dutch patients with dystrophinopathies and female carriers.

The DDD is a multicenter database of the **Duchenne Centre Netherlands (DCN)**, which captures a cohesive and extensive standardized dataset aligned to uniform core principles, like that of TREAT-NMD.

Aims

- **Facilitate the approach of eligible study participants**
- **Effective (re)use of patient and clinician reported data**
- **Ongoing study on epidemiology and natural history**

Methods

Design: The DDD holds a universal dictionary at the back-end that yields exchangeable information: By use of a system-independent information model that aligns to clinical guidelines, is constructed from detailed clinical models (ISO/TS 13972:2015), comprises data elements that are annotated with meta-information.
Data capture system: CastorEDC is used as electronic data capture solution and integration platform.
Inclusion: Patients may enroll in several different options (see figure 2). Only contact details are obligatory.
Access: Upon approval by the advisory board and the DDD board of directors, researchers may be granted access to data.

Data requests

Within the past two years, the DDD has been used to address seven enquiries received via TREAT-NMD, to inform and recruit eligible patients for four investigator-initiated studies, six pharmaceutical trials, and the conditional reimbursement of ataluren via the Dutch insured healthcare and for seven natural histories studies performed by DCN researchers.

Table 1: Overview of the registrants in the DDD.

Overview of the registrants in the DDD	Diagnosis								
	DMD		BMD		Intermediate		Carrier		Total
	number (n)	*age (years) mean±SD*	*number (n)*	*age (years) mean±SD*	*number (n)*	*age (years) mean±SD*	*number (n)*	*age (years) mean±SD*	*number (n)*
Total	495		162		18		29		704
Alive	316	(21.8 ± 9.4)	126	(40.4 ± 19.2)	14	(26.5 ± 12.1)	29	(46.4 ± 14.9)	485
Deceased	179		36		4		0		219
Genetic report available									
Total	406		128		17		13		564
DCN	207		33		8		2		250

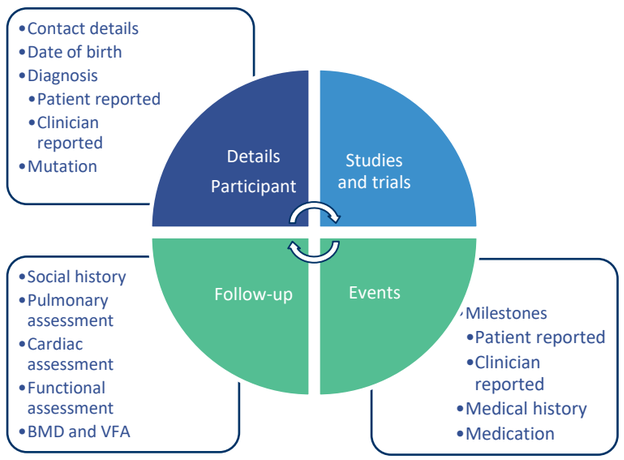

Figure 1: Overview of data collected in de DDD. The data is continuously updated. Event and follow-up data are obtained from patients having their outpatient visit(s) within the Radboud UMC or Leiden UMC.

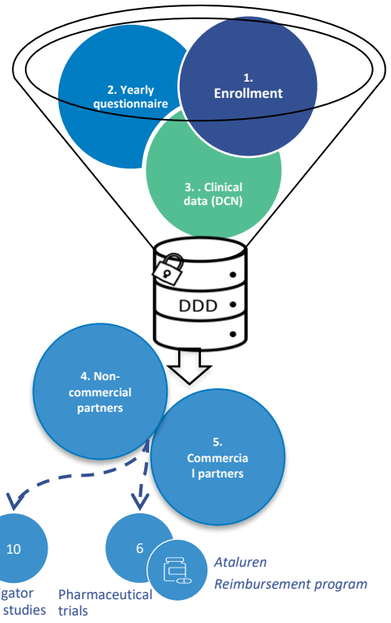

Figure 2: Patients may enroll in several different options. Only contact details are obligatory for **enrollment**. Registration option 2: **Yearly questionnaire** on disease milestones and use of medication. Registration option 3: Store **clinical data** acquired as part of regular care. Registration option 4 and 5 to consent for **exchange of coded data** with **non-commercial** and **commercial partners**, respectively. `

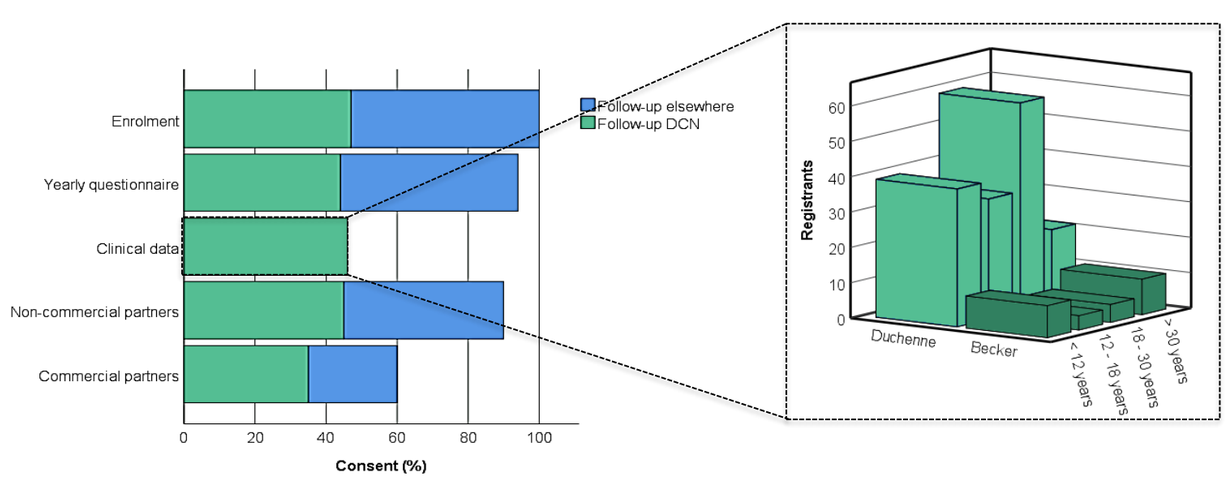

Figure 3: Percentage of patients who are alive and their consent per registration option. Clinical data can only be registered from patients having their outpatient visit within Radboud UMC or Leiden UMC for regular care.

Conclusion

The new structure of the DDD captures both patient reported and standardized healthcare data with modest burden for patients and clinicians.
The DDD has been proven useful to approach eligible participants for (clinical) trials, natural history studies, and for the conditional reimbursement of ataluren.

Future perspectives

Reduce of burden for patients and clinicians

- Implement e-consent to ease consent for patients
- Improve registration at the source

Enhance data use

- Integrate with the federated FAIR data queries
- Improve feedback to the patients, like linking with the Duchenne Data Platform
- Augment international collaboration

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