



Innovative FAIRification solution for a Rare Disease Patient-led Registry

Case Study: Duchenne Data Platform

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Introduction



Problem

Duchenne and Becker Muscular Dystrophy (DMD/BMD) data reuse prevented by silo mentality.



Solution

Make DMD data FAIR: Findable, Accessible, Interoperable and Reusable for humans and machines.



Outcome

DMD registry (Duchenne Data Platform) achieved FAIR Status within a year after a FAIRification process.

FAIR-in-a-Box

INPUT
Duchenne Data



EXTRACT



TRANSFORM



LOAD

OUTPUT
Duchenne FAIR Data



Patients

Store and access their health data through wearables or online



Clinicians

Access DMD FAIR (meta)data through an Authentication, Authorization Infrastructure (AAI)

Pathway for implementing FAIR principles



Set of Common Data Elements (CDEs)

Category	Element	Definition	Source
Demographics	Age	Age in years at the time of diagnosis	ICD-10
	Sex	Sex assigned at birth	ICD-10
	Ethnicity	Ethnicity	ICD-10
	Religion	Religion	ICD-10
Clinical	Diagnosis	Diagnosis code	ICD-10
	Severity	Severity of disease	ICD-10
	Prognosis	Prognosis of disease	ICD-10
	Outcome	Outcome of disease	ICD-10

Semantic (meta)data models



Methods

INITIATOR

Duchenne Parent Project, a Patient Organization in the Netherlands initiated the first FAIR-related project to transform Duchenne Data Platform (DDP), their patient-led registry, into a FAIR registry.

REGISTRY

DDP was built with interoperability in mind. Health Information collected are stored in 'personal datalockers' for easy access at all times by patients through wearables or online.

DATA

Variables to be collected correspond to the EU Minimal Set of Common Data Elements for Rare Diseases Registrations (CDEs) and Patient Reported Outcomes (PROs).

TEAM

DDP FAIR project required a multidisciplinary team of domain experts (Duchenne), FAIR Experts (Semantic Modeling, FAIR Infrastructure and Software Engineering), all supported by a FAIR project manager.

STANDARDS

Recommended International Standards by the European Joint Programme on Rare Diseases (EJPRD) were used. Ontologies were essential to annotate DDP data elements (e.g. HPO and NCIT).