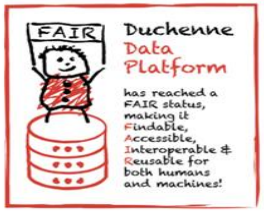

Innovative FAIRification solution for a Rare Disease Patient-led Registry

Case Study: Duchenne Data Platform

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SCAN FOR MORE INFO

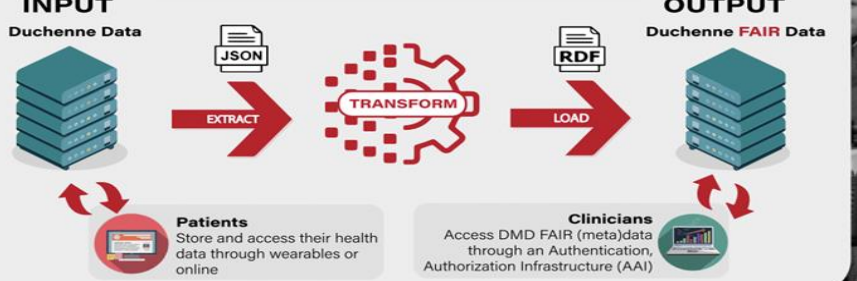
Introduction

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) achived FAIR Status within a year after a FAIRification process.

FAIR-in-a-Box



Pathway for implementing FAIR principles

Set of Common Data Elements (CDEs)

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).