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PER LA RICERCA FARMACOLOGICA
GIANNI BENZI
ONLUS

December 10th, 2021
Virtual meeting

XIV FORESIGHT TRAINING COURSE
The health emergency: regulatory crash and future perspectives

FAIR in practice: The Duchenne Data Platform

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World Duchenne Organization

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What is Duchenne Muscular Dystrophy?

Rare genetic disease defined by muscle weakness

Life expectancy: into early 30s with a % to late teens

Cause: mutations in the dystrophin gene

Incidence: 1 in 5000 newborn boys

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WORLD
DUCHENNE
ORGANIZATION
UPPMD

World Duchenne Organization

First FAIR Project

Duchenne Data Platform
The Netherlands

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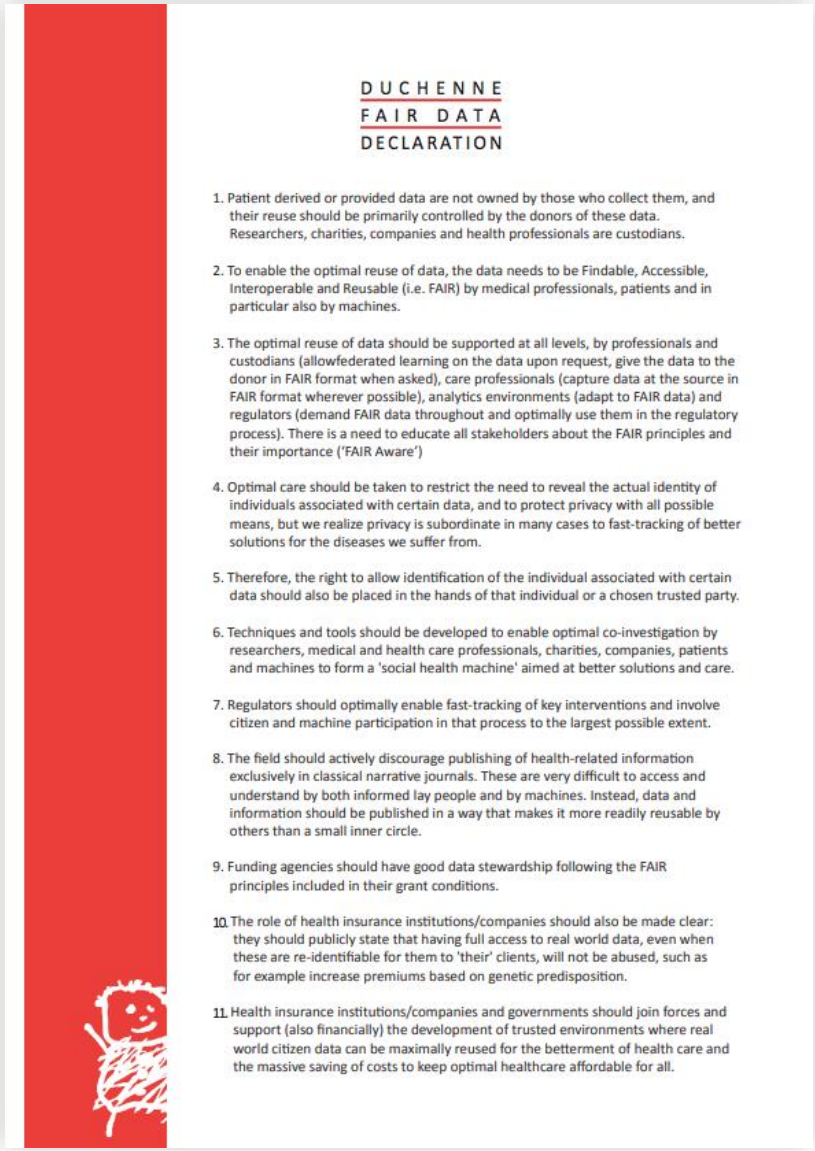
Duchenne Commitment to FAIR

2019
Initial workshop on Data Sharing for Duchenne
Published Duchenne FAIR Data Declaration

2020
Dedicated FAIR team in place
Started DDP FAIRification process

2021+
Further development for International Registrations
Roll-out to all other Duchenne data sources



DUCHENNE
FAIR DATA
DECLARATION

1. Patient derived or provided data are not owned by those who collect them, and their reuse should be primarily controlled by the donors of these data. Researchers, charities, companies and health professionals are custodians.

2. To enable the optimal reuse of data, the data needs to be Findable, Accessible, Interoperable and Reusable (i.e. FAIR) by medical professionals, patients and in particular also by machines.

3. The optimal reuse of data should be supported at all levels, by professionals and custodians (allowfederated learning on the data upon request, give the data to the donor in FAIR format when asked), care professionals (capture data at the source in FAIR format wherever possible), analytics environments (adapt to FAIR data) and regulators (demand FAIR data throughout and optimally use them in the regulatory process). There is a need to educate all stakeholders about the FAIR principles and their importance ('FAIR Aware')

4. Optimal care should be taken to restrict the need to reveal the actual identity of individuals associated with certain data, and to protect privacy with all possible means, but we realize privacy is subordinate in many cases to fast-tracking of better solutions for the diseases we suffer from.

5. Therefore, the right to allow identification of the individual associated with certain data should also be placed in the hands of that individual or a chosen trusted party.

6. Techniques and tools should be developed to enable optimal co-investigation by researchers, medical and health care professionals, charities, companies, patients and machines to form a 'social health machine' aimed at better solutions and care.

7. Regulators should optimally enable fast-tracking of key interventions and involve citizen and machine participation in that process to the largest possible extent.

8. The field should actively discourage publishing of health-related information exclusively in classical narrative journals. These are very difficult to access and understand by both informed lay people and by machines. Instead, data and information should be published in a way that makes it more readily reusable by others than a small inner circle.

9. Funding agencies should have good data stewardship following the FAIR principles included in their grant conditions.

10. The role of health insurance institutions/companies should also be made clear: they should publicly state that having full access to real world data, even when these are re-identifiable for them to 'their' clients, will not be abused, such as for example increase premiums based on genetic predisposition.

11. Health insurance institutions/companies and governments should join forces and support (also financially) the development of trusted environments where real world citizen data can be maximally reused for the betterment of health care and the massive saving of costs to keep optimal healthcare affordable for all.

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International FAIR Data Sharing for Duchenne

1st International FAIR meeting (2019)

36	Participants
6	Patient Organizations
6	Countries represented
10	Speakers & Facilitators
5	Clinicians
4	Pharmaceutical Companies
1	EU organization: EURORDIS

2nd International FAIR meeting (2021)

128	Participants
32	Patient Organizations
22	Countries represented
20	Speakers & Facilitators
19	Clinicians
8	Pharmaceutical Companies
3	EU organizations: EMA, EURORDIS, EJPRD

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International FAIR Data Sharing for Duchenne

1st Internation ... **eeting (2021)**

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Research Report

How Patient Organizations Can Drive FAIR Data Efforts to Facilitate Research and Health Care: A Report of the Virtual Second International Meeting on Duchenne Data Sharing, March 3, 2021

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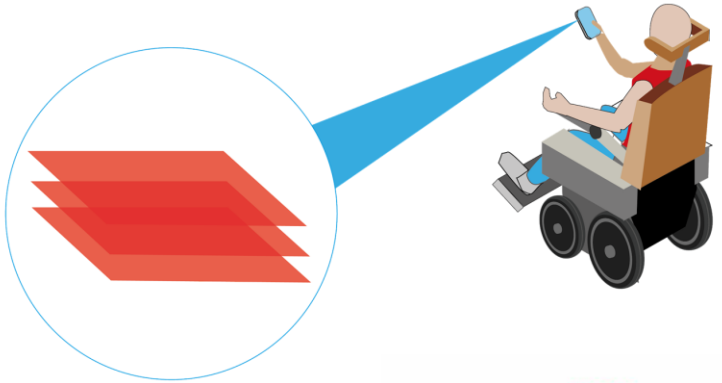
How did we activate our FAIR data?

The Registry

Duchenne Data Platform

- Online digital resource
- Built with interoperability in mind
- Access through smartphones (App) or computers
- Collects data relevant to patients
- Patients have their own virtual data ‘locker’
- Processing of personal data is based on consent
- Available for international registrations (In 3 languages)
- Managed by the Duchenne Parent Project, Netherlands

  www.duchenne.nl



Duchenne Parent Project

FAIR Data Systems

LUMC Leiden University Medical Center

Radboudumc

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The data

Patient Clinical Data

Most RD Common Data Elements (CDEs)

PROs

Use PROMs To measure the status of patients' health condition

Personal Data

Never accessed!

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Core FAIR team

- FAIR expertise: semantic modelling, FAIR infrastructure and software engineering

  FAIR Data Systems, Spain:
  Mark Wilkinson & team

  LUMC Biosemantics & FAIR data group, Netherlands:
  Marco Roos & team

- Advice, support & training

  Radboudumc, Netherlands:
  Peter-Bram ‘t Hoen & team

- Domain Experts

  Duchenne Parent Project, Netherlands:
  Elizabeth Vroom, Mirjam Franken

Bonus FAIR team

Core Team

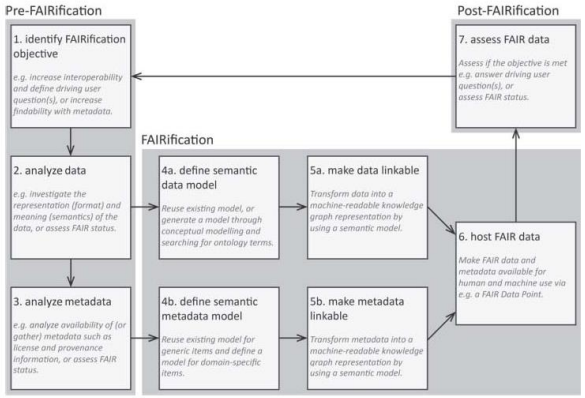
+

Bonus Team

EJPRD FAIR Stewards

EJPRD Experts

Used EJP-RD Recommended Standards



Pathway for implementing FAIR principles (Jacobsen et al.)

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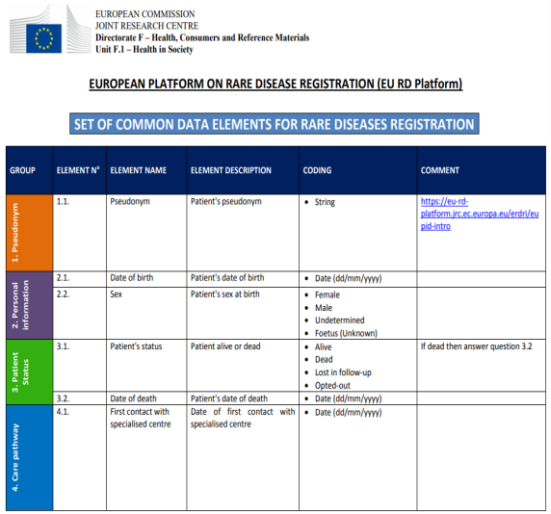

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JOINT RESEARCH CENTRE
Directorate F – Health, Consumers and Reference Materials
Unit F.1 – Health in Society

EUROPEAN PLATFORM ON RARE DISEASE REGISTRATION (EU RD Platform)

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	• String	https://eu-rd-platform.jrc.ec.europa.eu/erdri/eupid-intro
2. Personal information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	

Set of Common Data Elements (CDES) (EU RD Platform, JRC)

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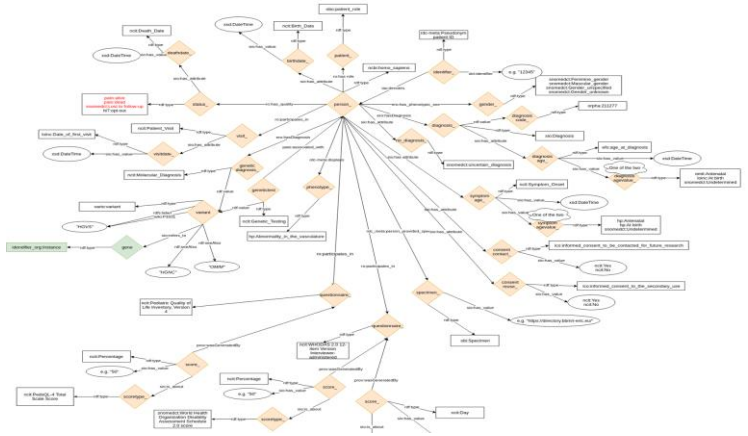

Semantic (meta)data models (CDE-semantic-model)

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FAIR approach: Retrospective

- FAIR through a series of transformations
- Adding a FAIR layer on top of an existing infrastructure
- Registry already existed and well in use
- No or minimal changes to the original system
- Data stored as JSON files
- Retrieved through API calls

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FAIR transformation: Solution found and deployed!

"CDE in a Box"

- NEW FAIR transformation tool, specially developed for this project
- Ready - all the hard work has been done!
- Simple format - data to be FAIRified are organised in a CSV file
- Sustainable as based on existing international standards
- Automated process with nightly updates
- Available to others to test and use!

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CDE in a Box

INPUT

Duchenne Data

1

JSON

Extract

2

Transform

3

RDF

Load

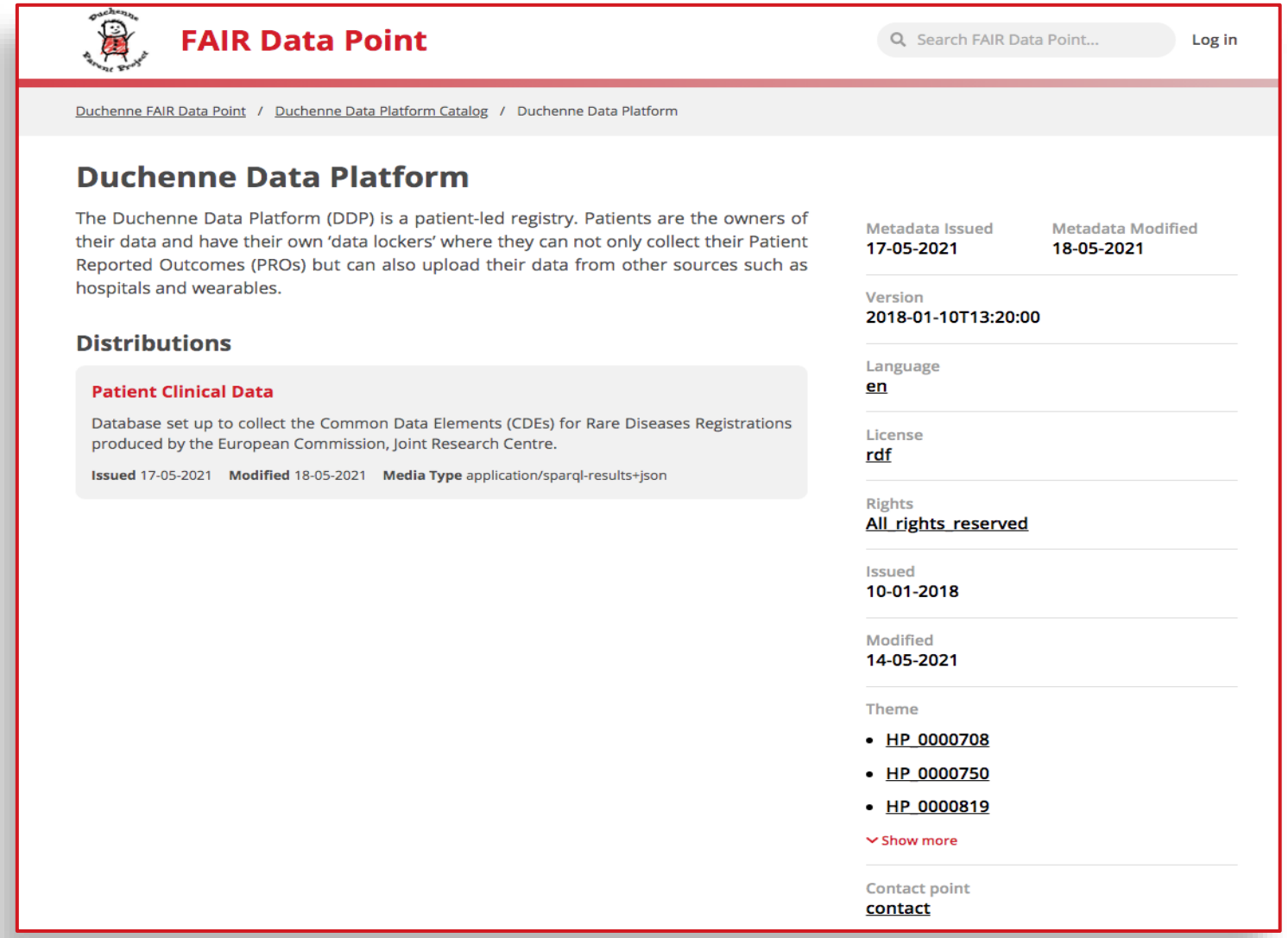
OUTPUT

Duchenne FAIR Data

Patients
Store and access their health data online

Clinicians
Access FAIR data though SPARQL queries

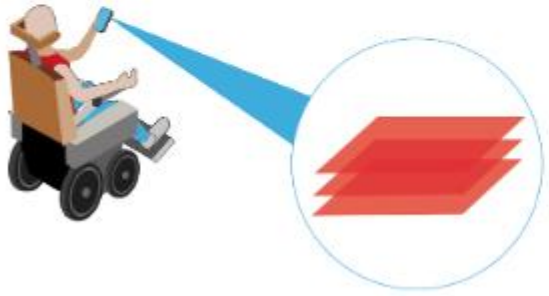
FAIR Data Point - Deployed!

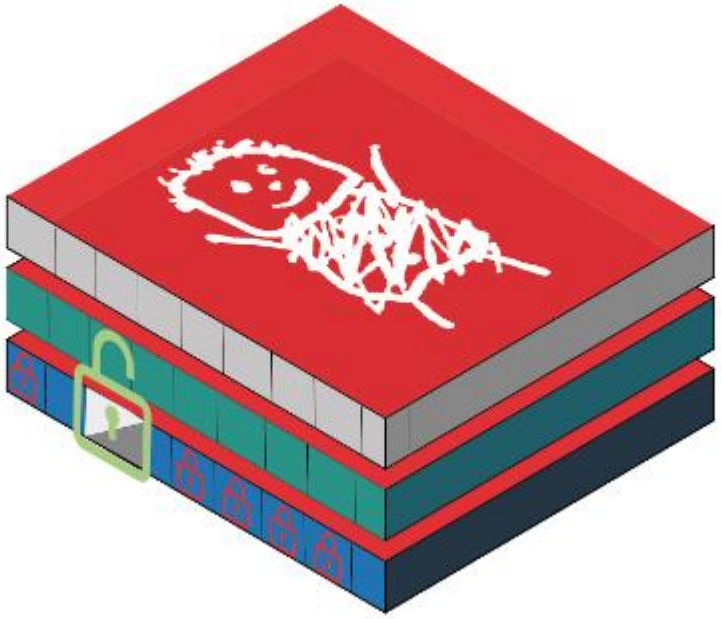


View metadata

Access FAIR data

DPP is FAIR Registry!





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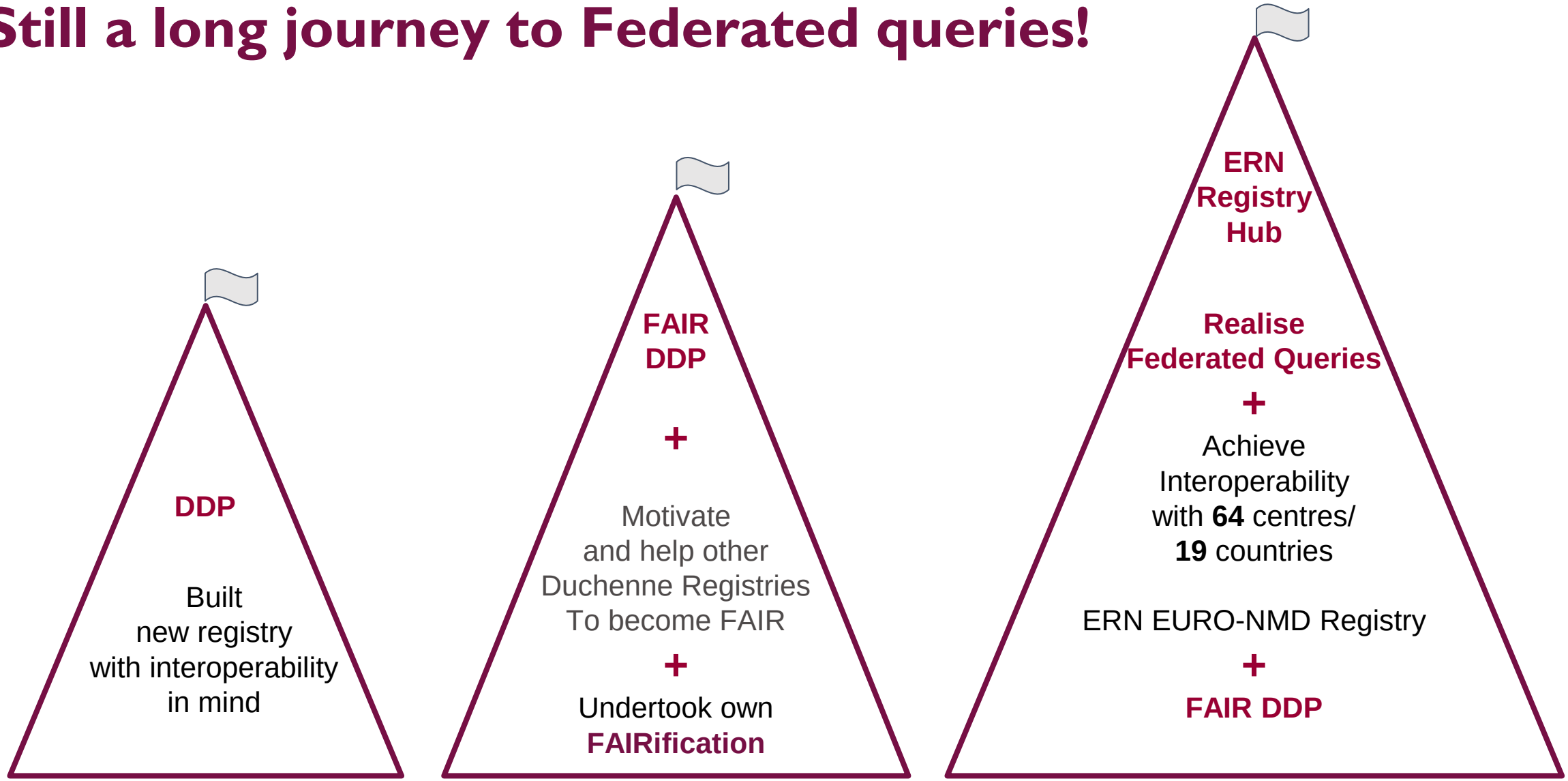
Data
Platform

Still a long journey to Federated queries!



Take home messages

1. Patient Organizations such at Duchenne stand their support to drive change
2. FAIR is FAIR for all stakeholders as all stand to benefit
3. FAIR enables data reuse at scale by machines
4. Implementation is a multidisciplinary process
5. Dialogue is key to building a solid ecosystem for data ‘visiting’
6. Find out more about FAIR www.go-fair.org

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7. Data Stewards at EJP-RD
8. EURO-NMD

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Thank you!

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