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FISMA-governed registration at the source in Duchenne-Becker Biobank

FAIR Real-world data for multiple purposes

INTRODUCTION,
Duchenne and Becker muscular dystrophy are rare, progressive multifactorial muscular diseases, with often a fatal outcome. Thus, high-quality, interchangeable, and reusable real-world data (RWD) from a representative DBMD population are essential to improve clinical trials, understand disease progression, and advance biomarker discovery. Duchenne Center Netherlands developed FISMA (Framework for Information Specification, Modelling and Architecture) to curate all clinical observations and biomaterials at the source, which are captured in a structured, semantically interoperable way directly within routine care.

AIM
To implement an inherently FAIR approach, that enables collection of high-quality, curated clinical & biomaterial data from LUMC's Electronic Health Record (HiX) and Biobank Information System (BIMS/Sample Navigator), where FISMA has been operational since 2020.

CONCLUSION
Data registration through FISMA principles allows collection of interoperable, reusable sample and clinical data (RWD) for all consented DMD/BMD patients in the Biobank (2020–2025).

Figure 1A. FISMA governs every step of RWD collection, from implementation to distribution.

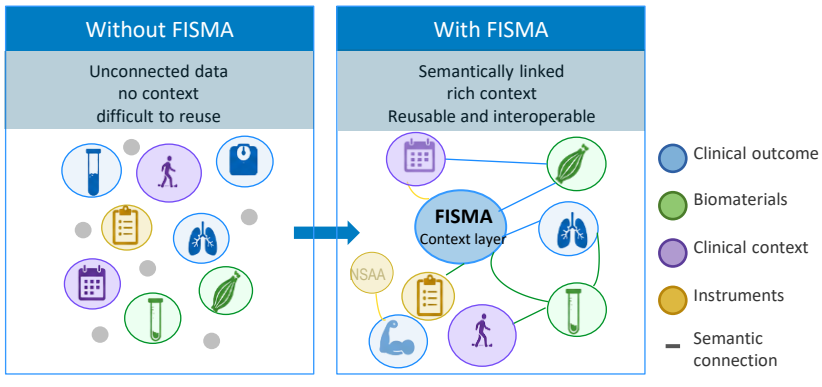

Figure 1B. From unconnected data to data richly embedded in its context with FISMA.

<50% / 50-75% / 75-100%	Becker muscular dystrophy						Duchenne muscular dystrophy					
	Informed consent		Biomaterials, yearly			one time	Informed consent		Biomaterials, yearly			one time
YEAR	total	new	Blood	RNA	Urine	DNA	total	new	Blood	RNA	Urine	DNA
PRE 2020	17						58					
2020	22	(+ 5)	12	11	12		86	(+ 28)	43	42	49	
2021	25	(+ 3)	13	13	12		107	(+ 21)	63	60	62	
2022	32	(+ 7)	18	13	16		123	(+ 16)	93	62	86	
2023	36	(+ 4)	25	1	22		131	(+ 8)	85	2	89	
2024	39	(+ 3)	22		21		141	(+ 10)	101		97	
2025	40	(+ 1)	21		15		146	(+ 5)	100		74	
Total	40					30	146					109

<50% / 50-75% / 75-100%	Becker muscular dystrophy						Duchenne muscular dystrophy					
	Informed consent		Clinical data				Informed consent		Clinical data			
YEAR	total	new	Physio	Clinic	lab	Med	total	new	Physio	Clinic	lab	Med
PRE 2020	17						58					
2020	22	(+ 5)	13		15	18	86	(+ 28)	51	5	70	86
2021	25	(+ 3)	18	7	20	26	107	(+ 21)	76	56	91	100
2022	32	(+ 7)	21	7	22	21	123	(+ 16)	89	29	99	104
2023	36	(+ 4)	25	10	29	24	131	(+ 8)	91	60	101	106
2024	39	(+ 3)	16	8	22	18	141	(+ 10)	85	83	107	110
2025	40	(+ 1)	26	11	18	23	146	(+ 5)	98	98	107	110
Total	40						146					

FIGURE LEGEND - coverage per year 2020-2025
Comparison of biomaterials (at top) and clinical dataset (below) completeness between Becker (left) and Duchenne (right) muscular dystrophy cohorts. The heat-map shading indicates the percentage of available data per category (blood, RNA, urine, DNA, **physio**therapy, **clinic**al, **lab**oratory, and **med**ication records), highlighting differences in sample availability and consent across years.

RESULTS
As expected, fewer individuals with Becker were included than those with Duchenne muscular dystrophy. For biomaterials, 50–75% of patients have available blood and urine samples, and over 75% have DNA samples. Clinical data, specifically physiotherapy assessments, medication records, and laboratory values from the electronic patient dossier, are available for approximately 50–90% of included patients.

As part of our analysis, we found that Becker and Duchenne muscular dystrophy patients carry an average of 5.6 and 8.0 specialist-assigned diagnoses, respectively, reflecting a level of diagnostic complexity that is rarely captured in traditional research proposals. Together, these findings showcase the depth, completeness, and real-world clinical richness of the dataset.

1 PROBLEM
Data not useable
Silos, different sources, not interoperable nor findable

2 SOLUTION
FISMA (inherently FAIR)
Semantic layer, clinical context and provenance

3 IMPLEMENTATION
FISMA in EPD & BIMS
Registration at the source
Interoperable

4 IMPACT
Biomarker study
New insights, better trials

THE TAKEAWAY
Curation at the source converts routine care data into ready-to-use resources that directly accelerate biomarker research.

More information
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