InGene: a digital platform for deep phenotyping in neuromuscular diseases

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The Health360 Framework

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HEALTH360

Framework applicativo
In cloud

Developed in the projects:

- Medical360 (Regional Call 2009 for Support to industrial research and experimental development projects between SMEs and research organizations in the field of health) 2013-2015
- InGene (Regional Health Research Call 2014) 2017-2019
- InGene 2.0 (Regional Health Research Call 2018) 2020-2024
- BrAId (Regional Health Research Call 2018) 2021-2024
- Telos (Regional Health Rese arch Call 2018) 2021-2024

Regione Toscana

InGene

braid

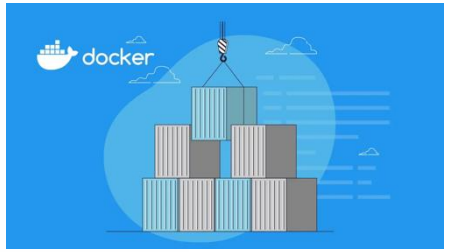
Actually used in projects:

- DiGiMe (European Health and Digital Executive Agency)
- BPReg (PNRR Italian Ministry of Health)

Goal: to implement the 4P (+1) medicine

- Preventive
- Predictive
- Partecipatory
- Personalized…
- …and Precise

Modularity & flexibility

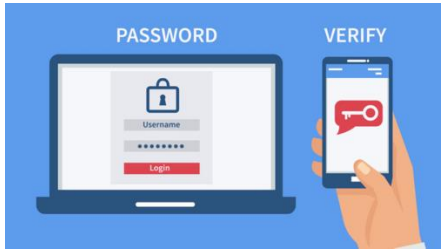


Each module is built on a separate environment

Role Based Access Control

Each user only accesses the data of their own organisation
But the analysis is perfomed on the entire dataset

2F Authentication



Required by local laws to process genetic data

the Domain: Neuromuscular Diseases

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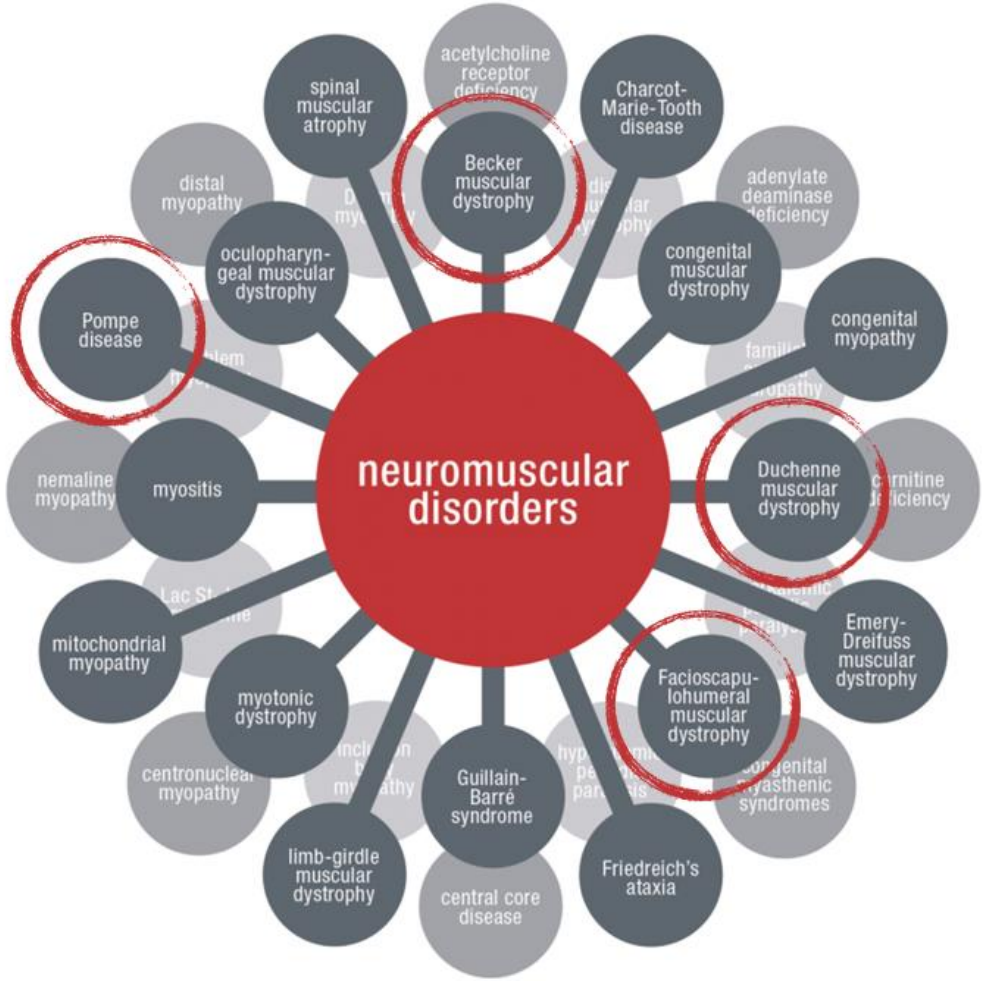

The InGene and InGene 2.0 projects

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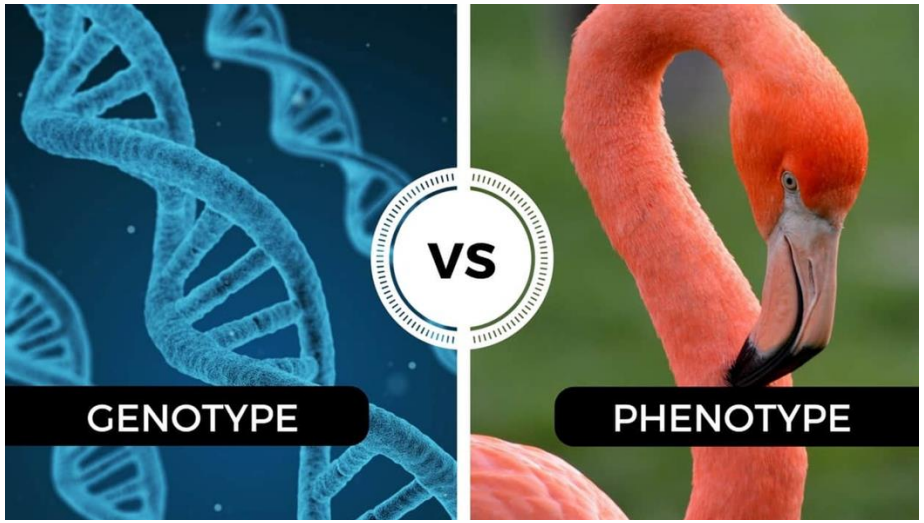

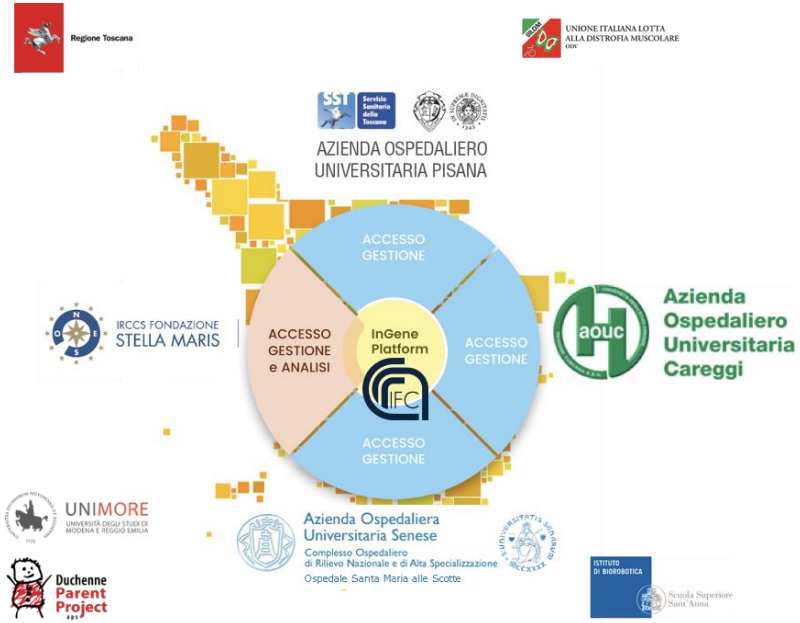

InGene

The projects propose to simultaneously use different data (**clinical, bioinformatics, biomechanical…**) in order to make the **genotype/phenotype correlation** more reliable in a large cohort of patients affected by **neuromuscular diseases**, studied using new generation techniques, and thus help the clinician in formulating treatment plans, monitoring the pathology and, therefore, in **patient care**.

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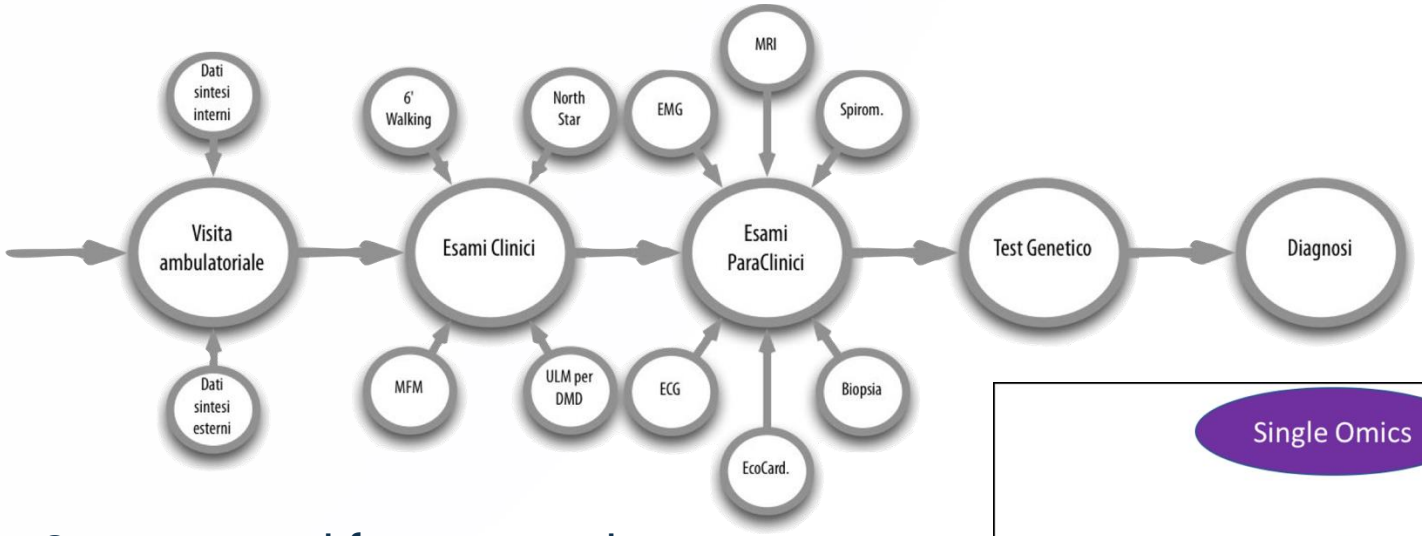
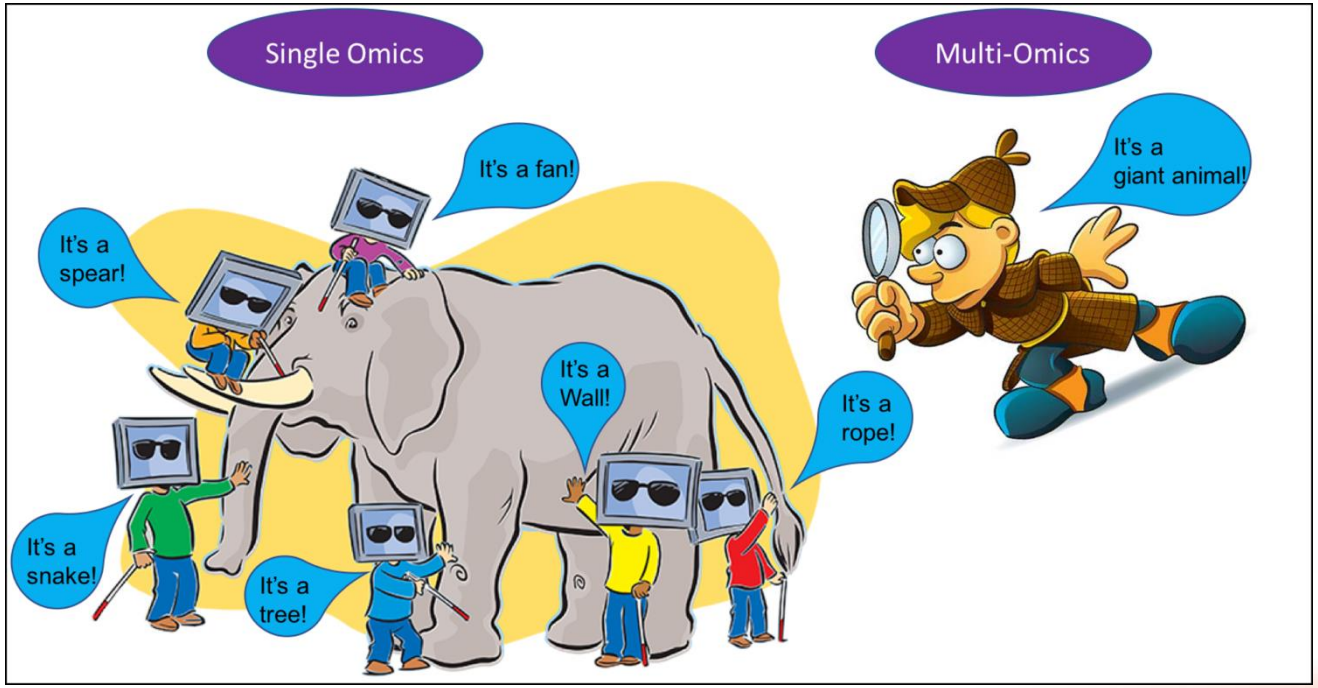
The AIM of InGene Platform



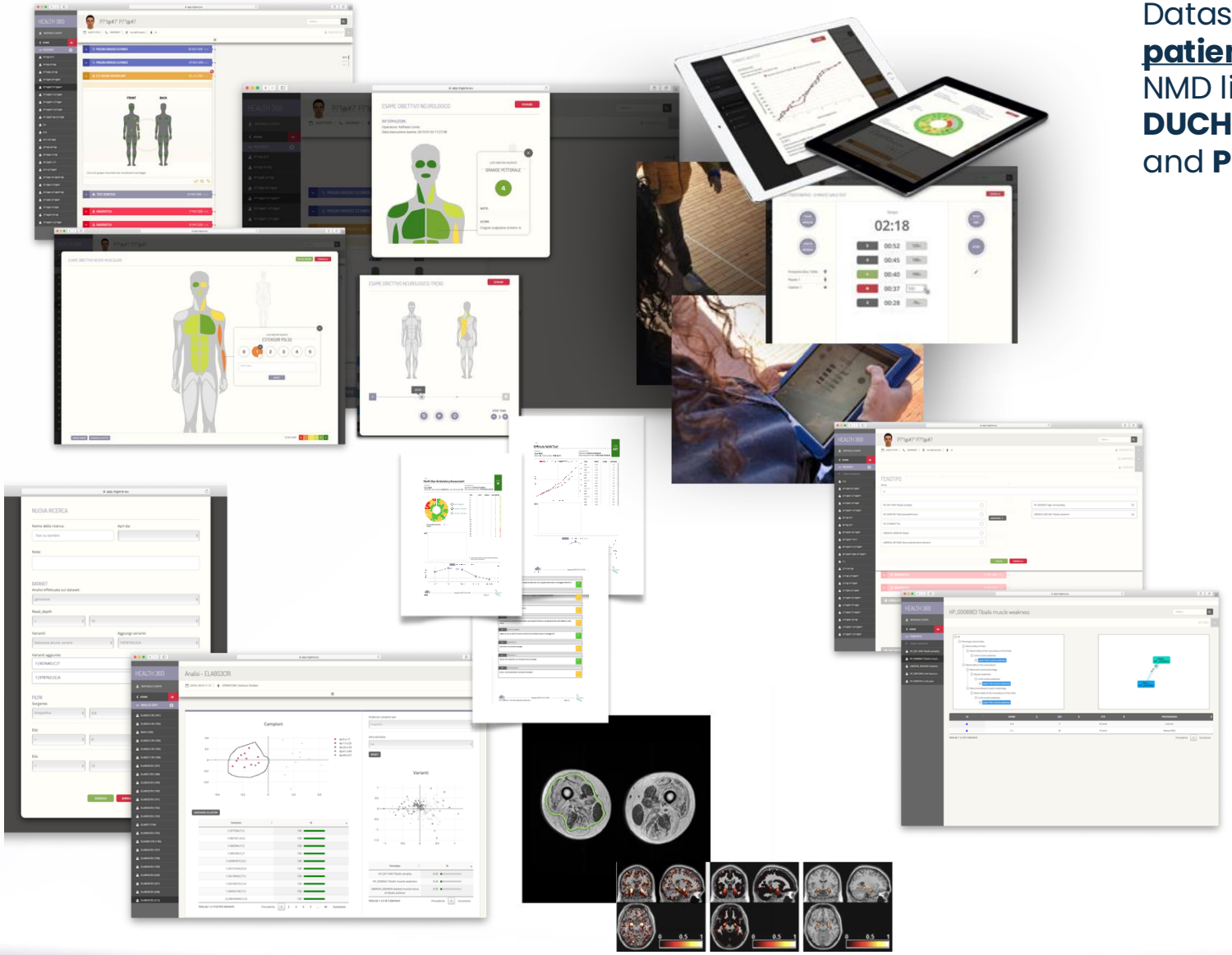
Support tool for research and aid for diagnosis and therapy

Integration of multiparametric and multimodal data

A Holistic approach



The InGene Platform: results



Dataset involving **500 patients** affected by NMD like **FSHD**, **DUCHENNE**, **BECKER** and **POMPE**

Modular SaaS Platform

Wrap Up

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- A holistic approach to Neuromuscular Diseases
- Integration of high-dimensional, heterogeneous datasets
- Deep data integration to improve Phenotype-Genotype correlation
- Technology build for a better GDPR compliance

The CNR Team

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Thank You!!

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