



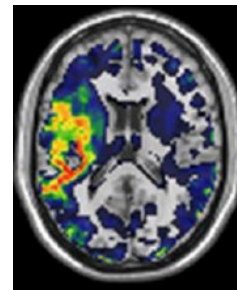
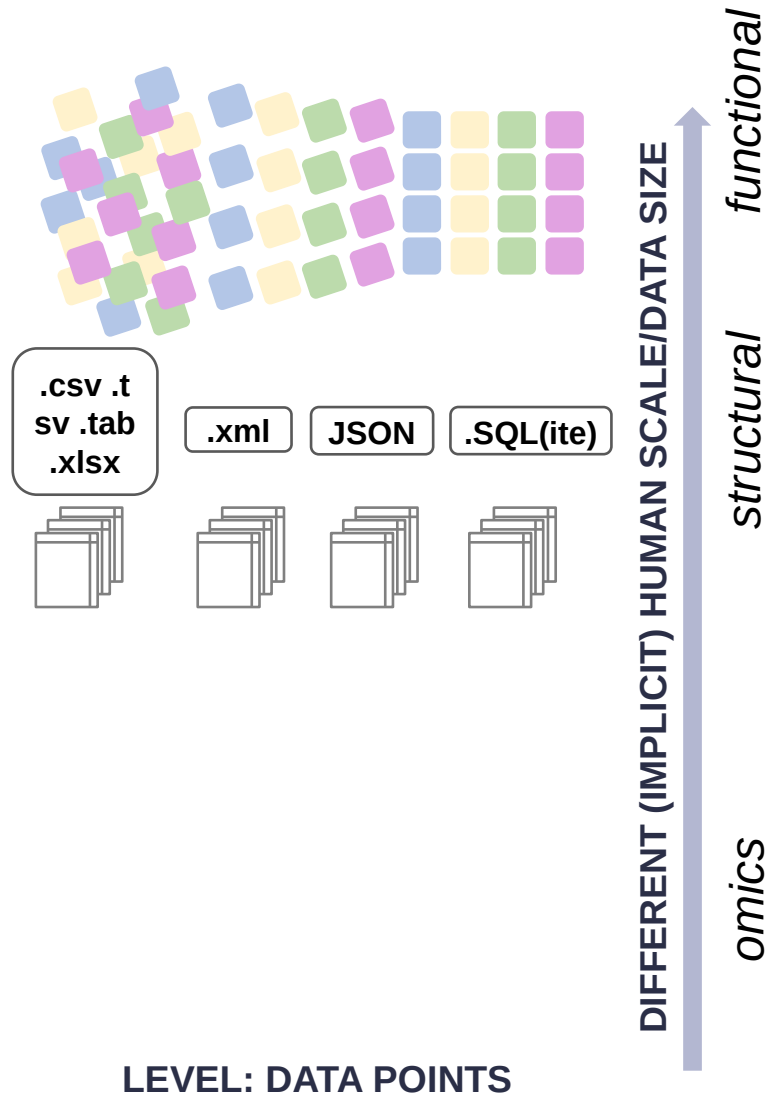
Health data integration and exploitation

Sandrine Muller, Msc. Eng., PhD
Chaire de Professeur Junior, AI in health
Laboratoire d'Informatique de Grenoble (LIG)
sandrine.muller@univ-Grenoble-alpes.fr

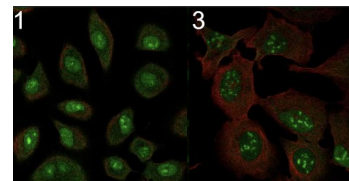
Health data types

HETEROGENOUS DATA SOURCES CURATION AND STRUCTURATION

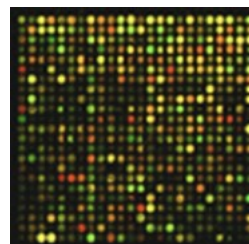
KNOWLEDGE REPRESENTATION GRAPHS & ONTOLOGIES



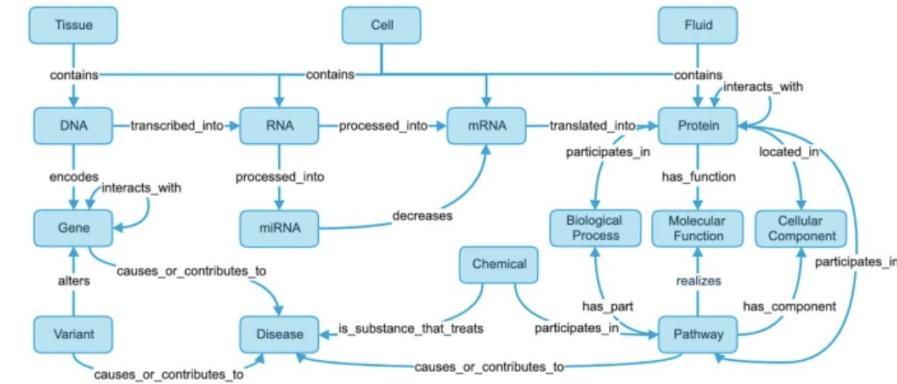
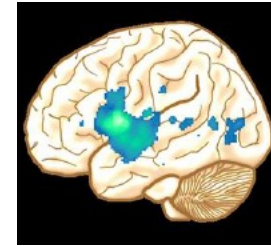
Muller *et al.*; Curr. Top. in Med. Chem.; 2020



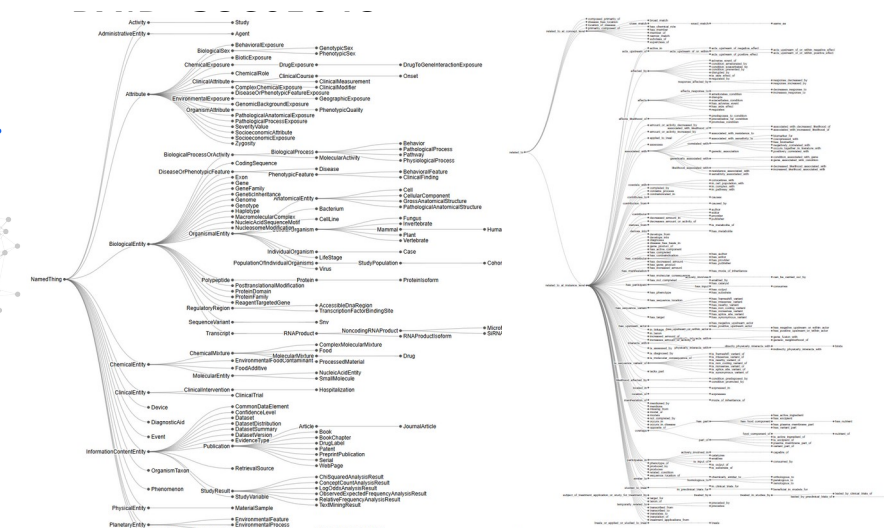
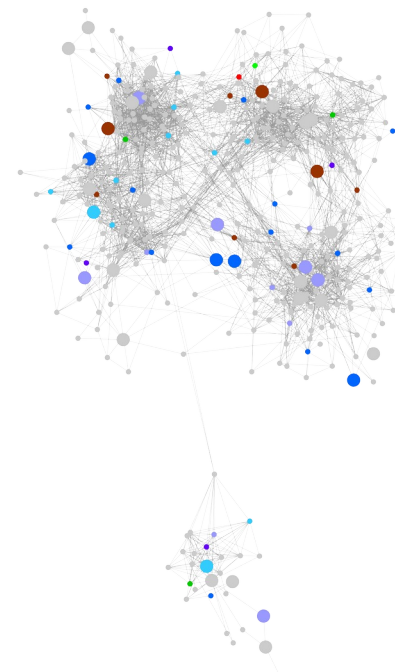
proteinatlas.org



yourgenome.org



Callahan *et al.*; Scientific Data; 2024.



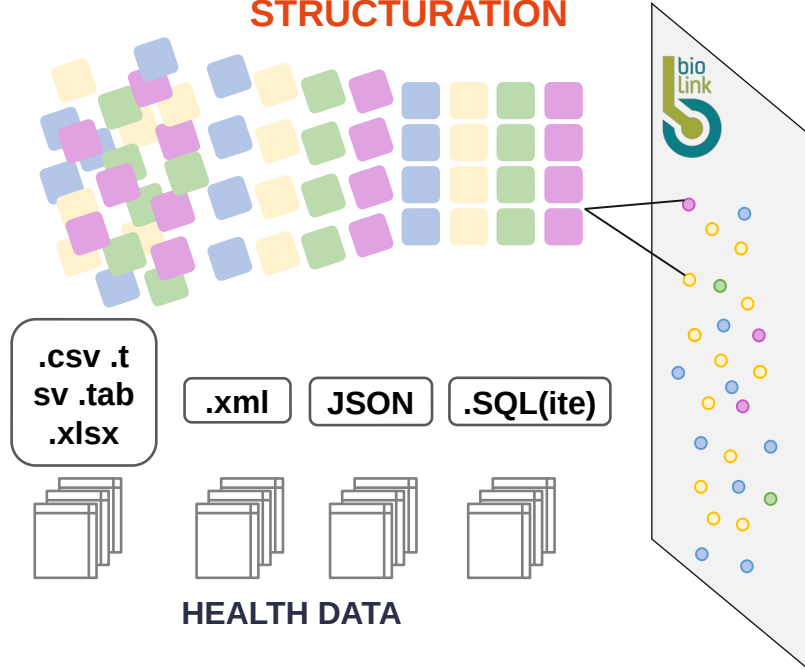
Unni *et al.*; Clin. Transl. Sc.; 2022.

PMID: 36125173

LEVEL: SUMMARY STATISTICS - PATTERNS
HUMAN EXPERTISE – CURATED DATA

Health data integration and exploitation

HETEROGENEOUS DATA CURATION AND STRUCTURATION

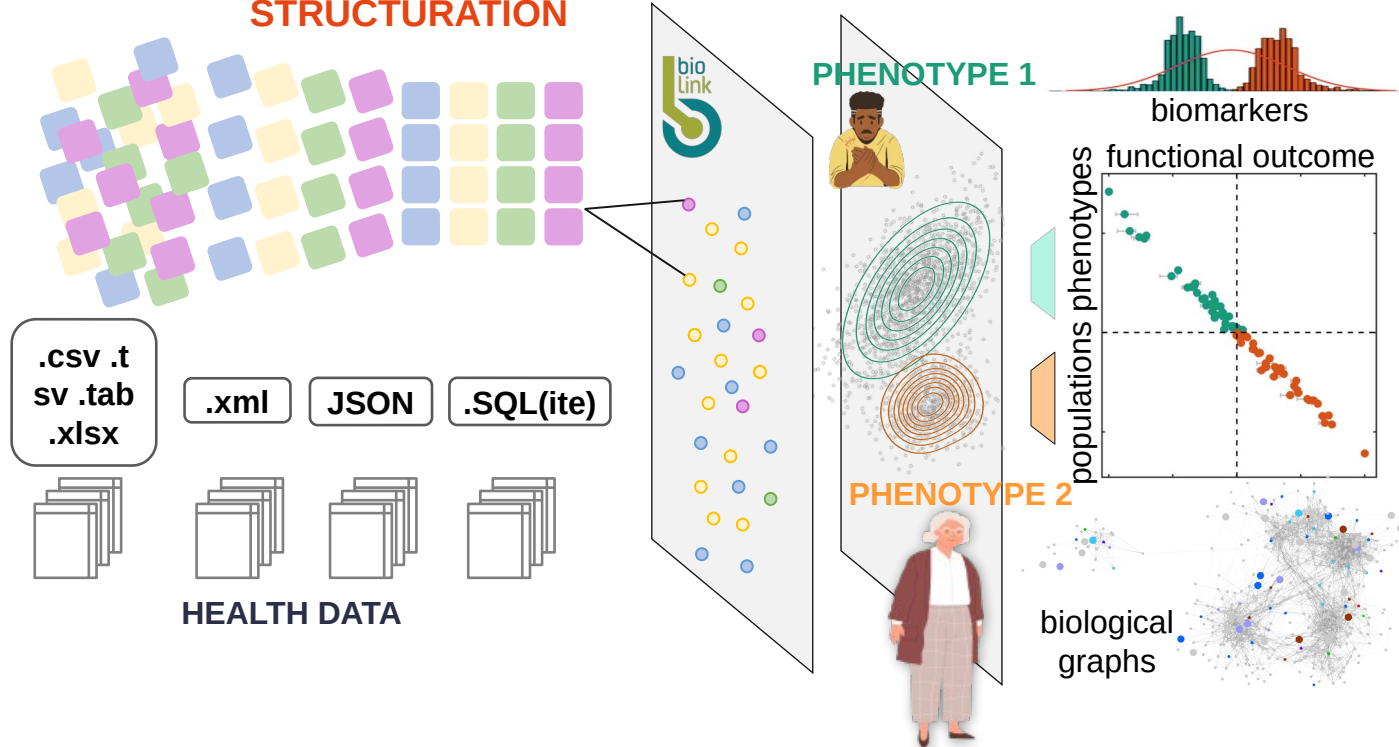


Produce knowledge
Structuring
Sharing (FAIR)

Health data integration and exploitation

HETEROGENEOUS DATA CURATION AND STRUCTURATION

MODELIZATION



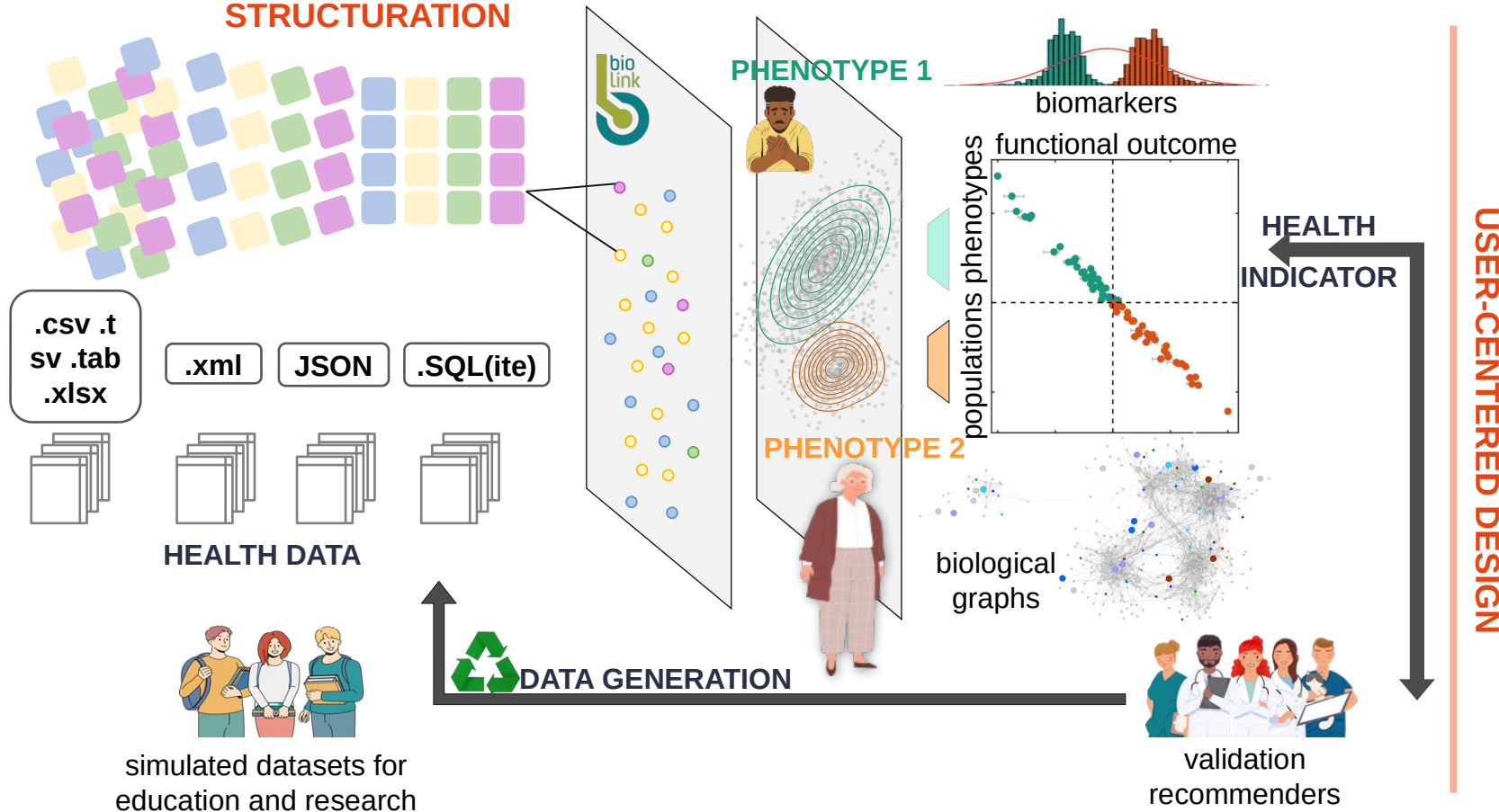
Produce knowledge
Structuring
Sharing (FAIR)

Phenotyping
Biomarkers
Biological graphs

Health data integration and exploitation

HETEROGENEOUS DATA CURATION AND STRUCTURATION

MODELIZATION



Produce knowledge
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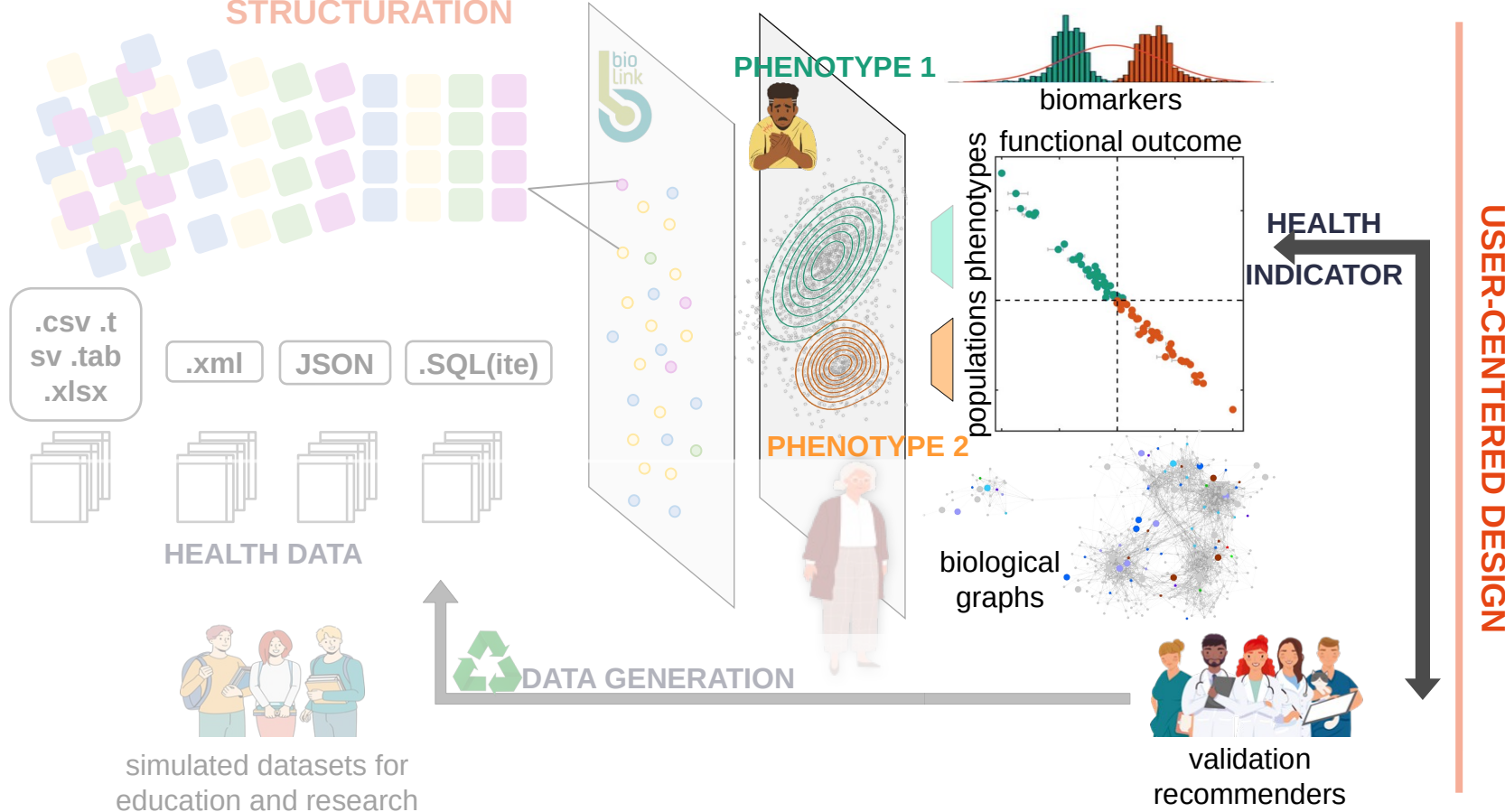
Prevention
Education and training
User-centered design

AUTOMATED, SECURE, AND REPRODUCIBLE PIPELINES

Health data integration and exploitation

HETEROGENEOUS DATA CURATION AND STRUCTURATION

MODELIZATION



AUTOMATED, SECURE, AND REPRODUCIBLE PIPELINES

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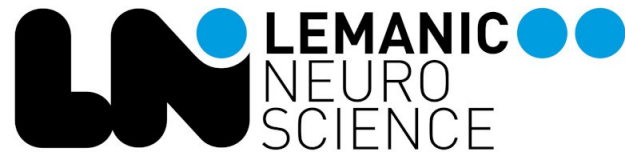
Prevention
Education and training
User-centered design

**Transparent, intuitive,
and customizable AI to
answers biomedical
questions**

Encoding human behaviors and researcher needs

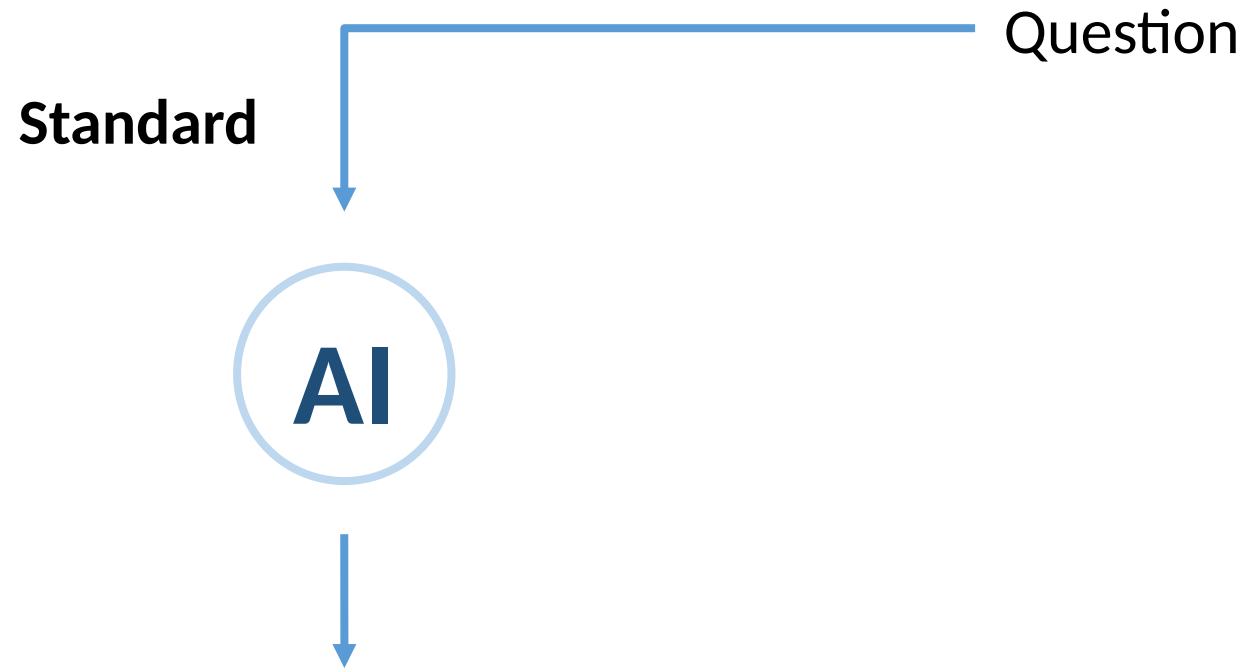


- PhD. Neuroimaging, methodology (Lemanic Neuroscience Doctoral School, Switzerland)
- Master of Engineering in
 - Electronics, Telecommunications and Informatics (CPE Lyon)
 - Neuroengineering (EPFL)

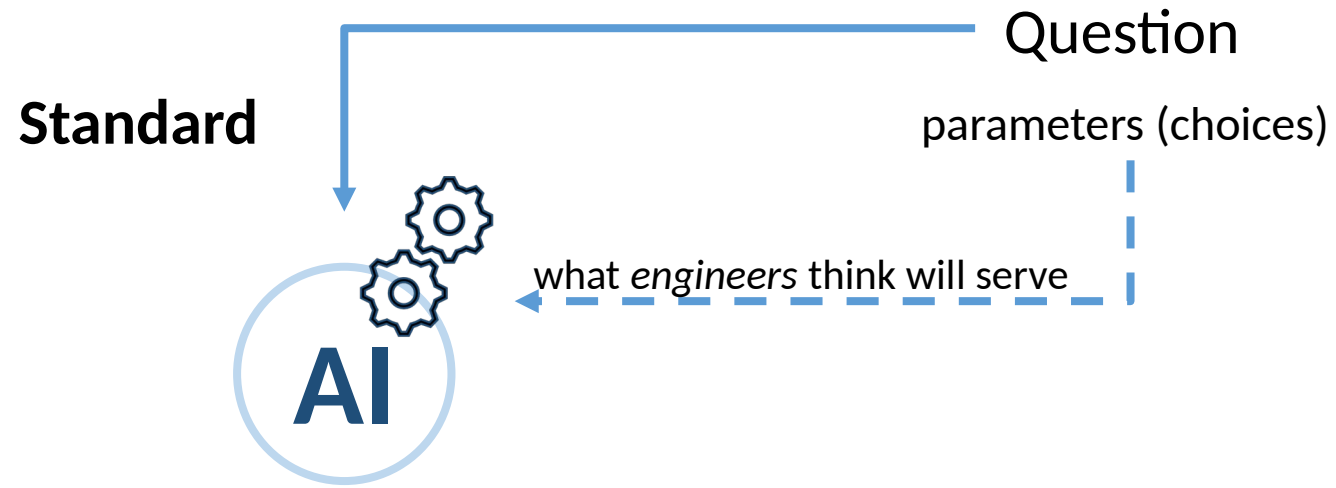


The problem is accessibility

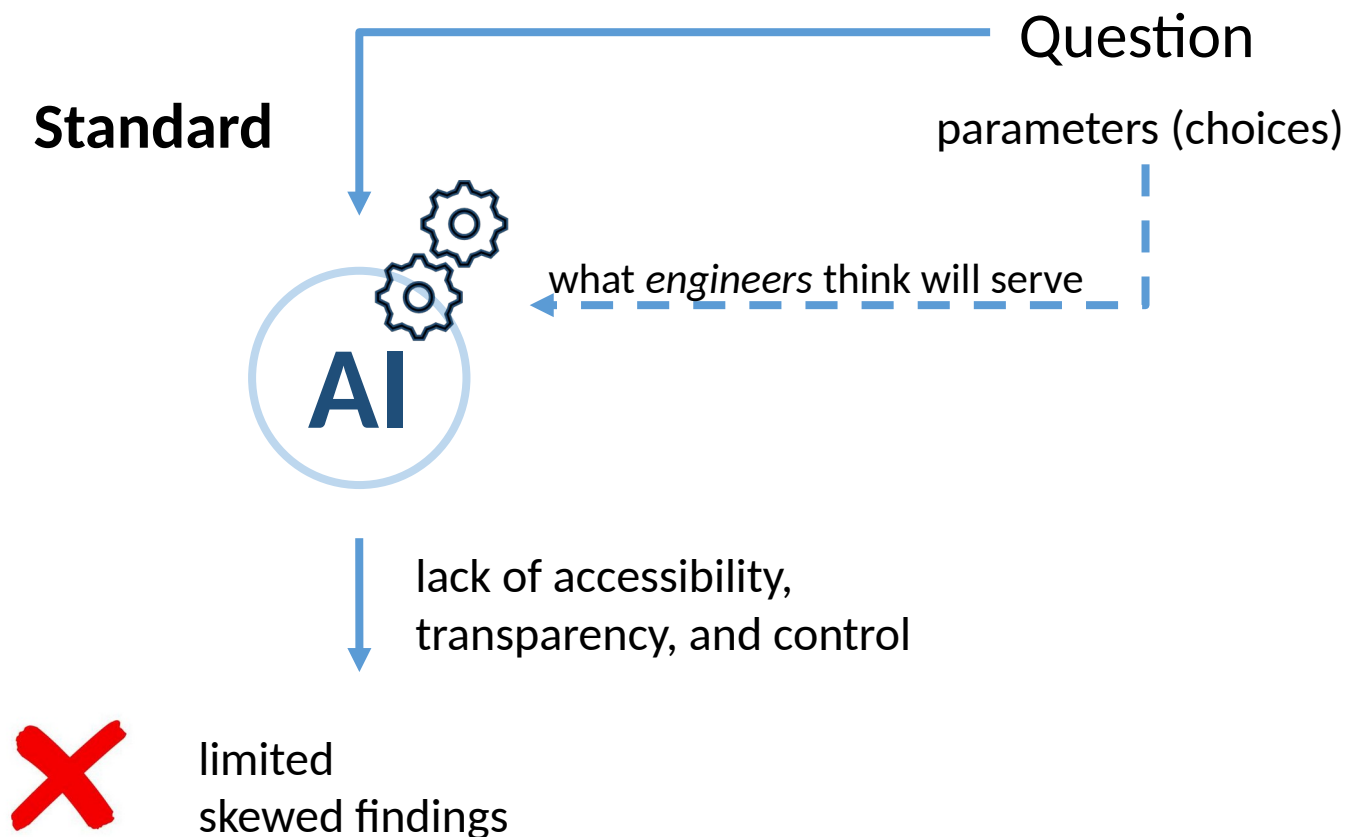
The problem is accessibility



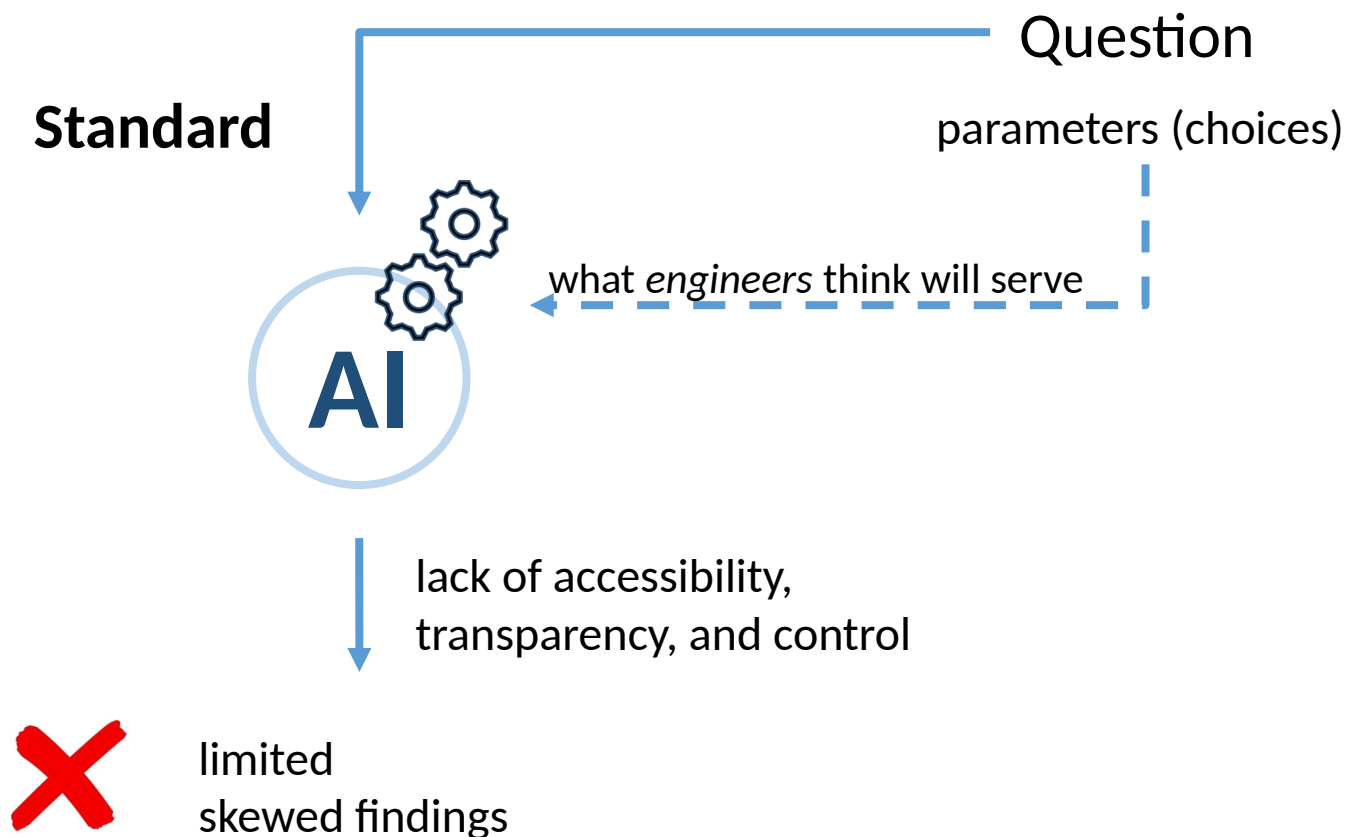
The problem is accessibility



The problem is accessibility

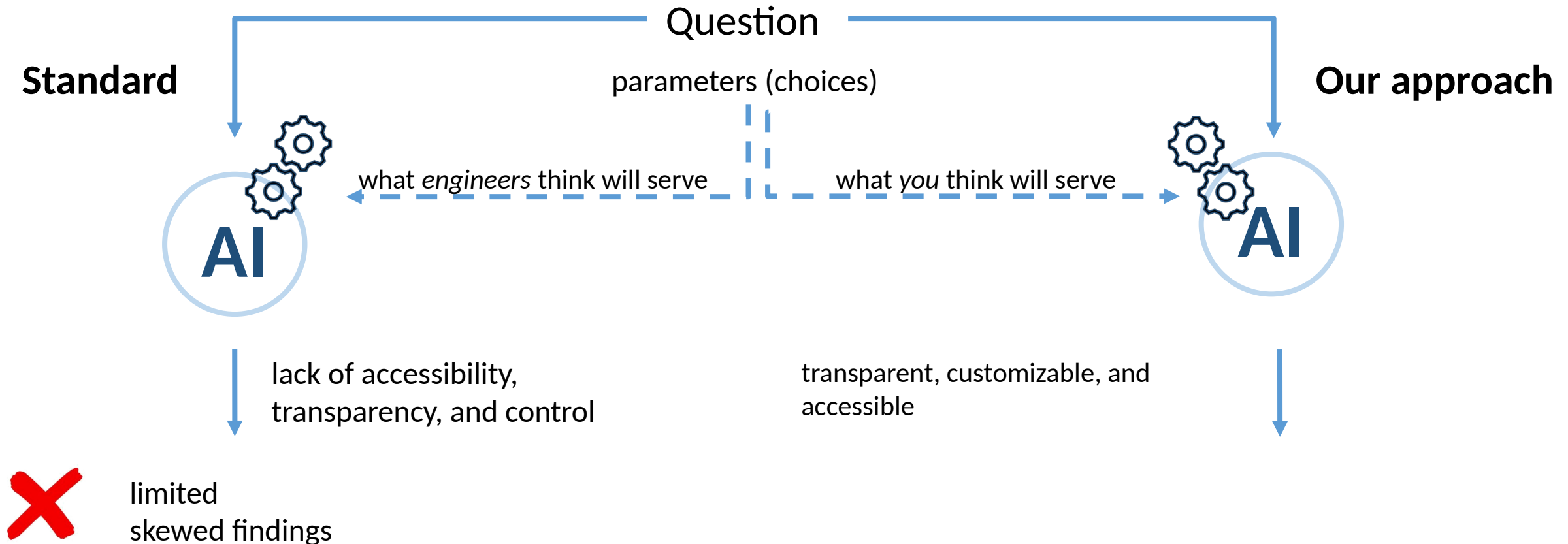


Tuning AI systems from user preferences



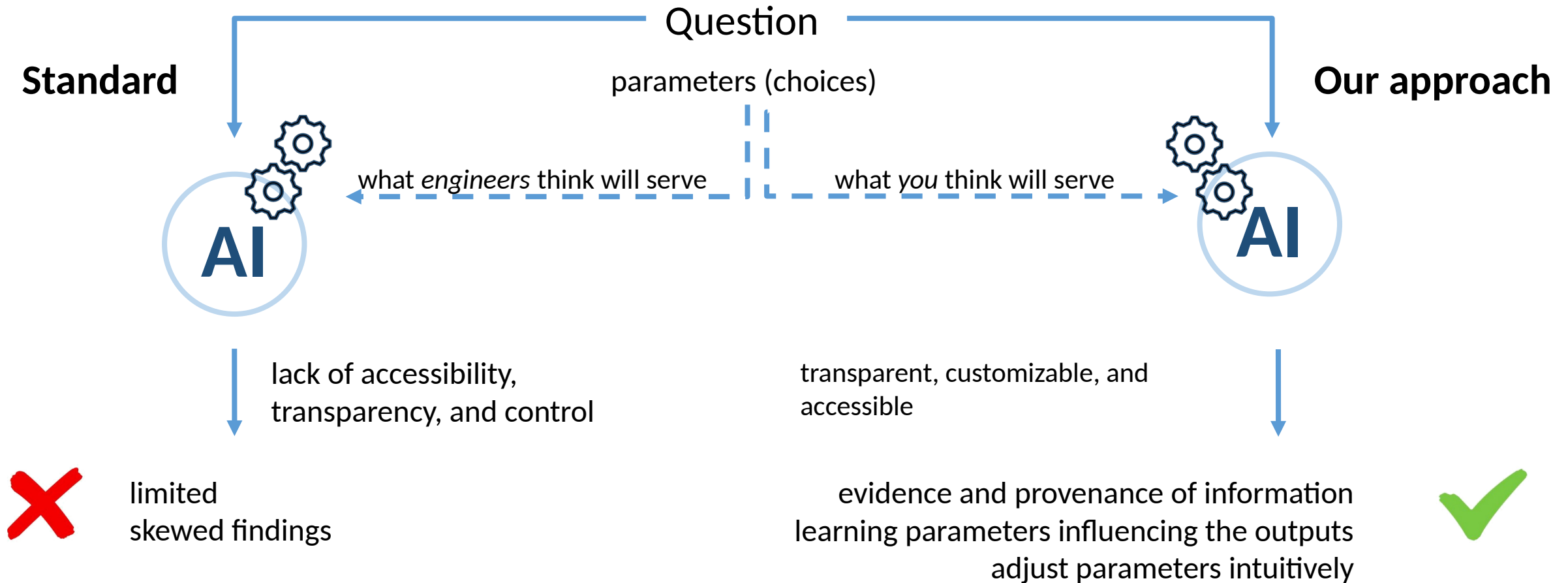
Tuning AI systems from user preferences

*Science **by** and **for** scientists*



Tuning AI systems from user preferences

*Science **by** and **for** scientists*



The FSHD1 use case

Collaboration USMB and UCL

The FSHD1 use case



**Christophe
Hourdé, PhD,
LBFA, USMB, FR**



**Julie
Dumonceaux,
PhD, UCL, UK**

The FSHD1 use case



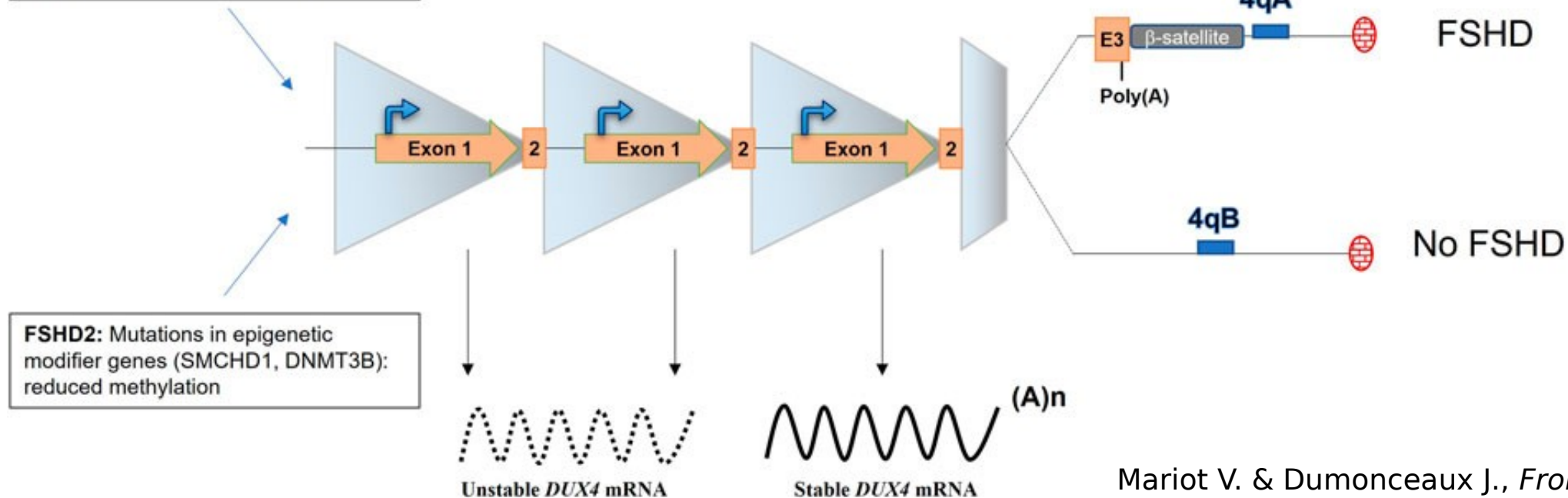
Christophe
Hourdé, PhD,
LBFA, USMB, FR



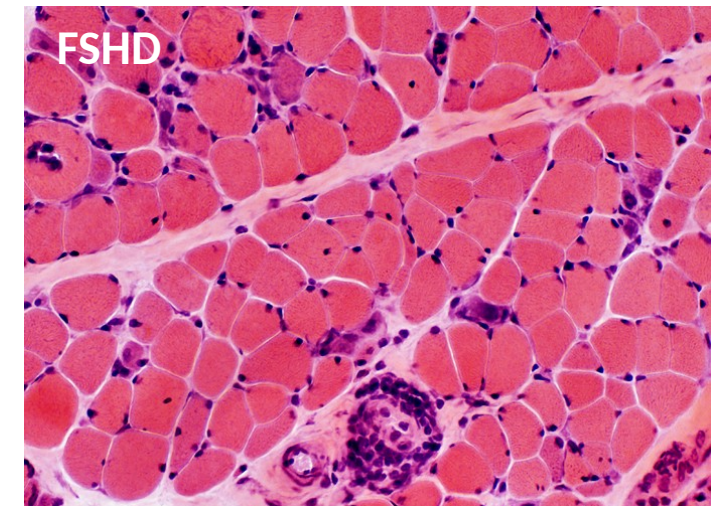
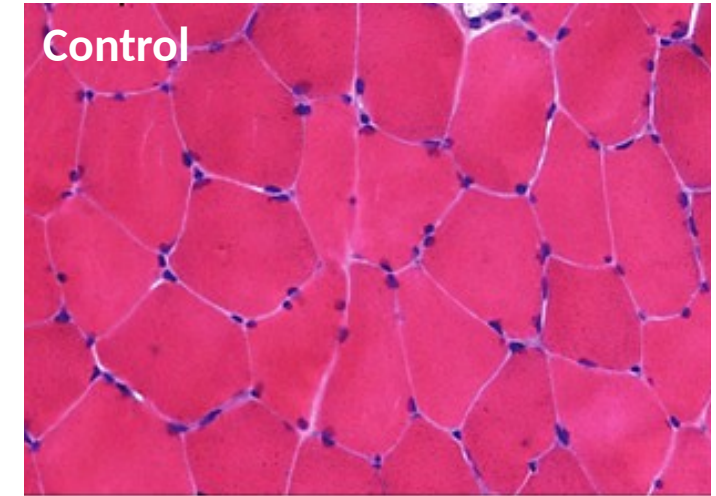
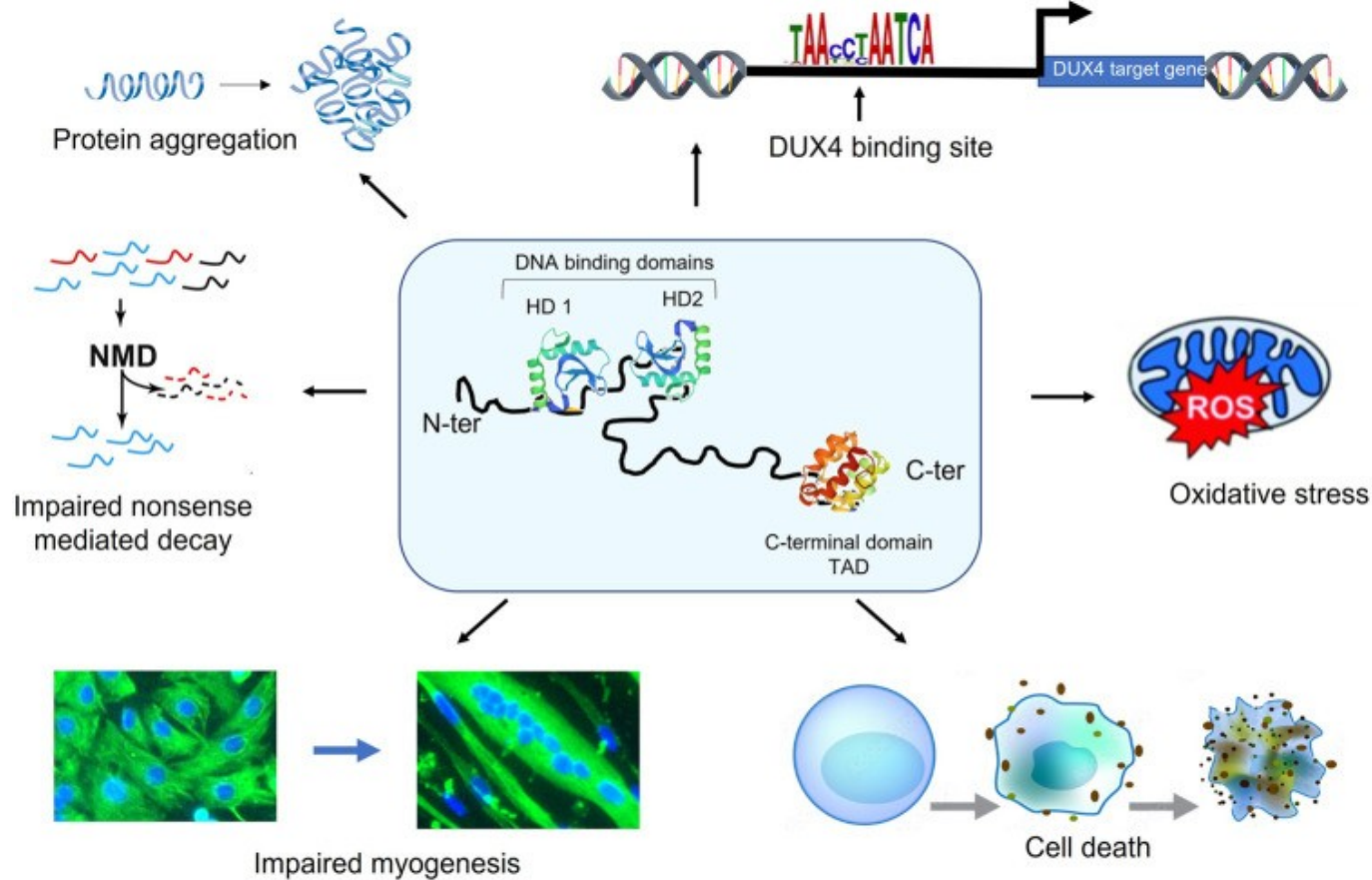
Julie
Dumonceaux,
PhD, UCL, UK

FSHD1: Contraction of the D4Z4 array:
hypomethylation of the D4Z4 array

- FSHD is the third most common type of muscular dystrophy. Currently no preventative treatments or cures.
- Primarily affected muscles are the face, shoulders, upper arms, but not limited to these muscle groups.
- Hypomethylation of D4Z4 repeats in chromosome 4 results in production of toxic DUX4 protein.



DUX4 toxicity and damage in muscles in FSHD



Banerji CRS & Zammit PS., *EMBO Mol Med.* (2021)

ariot V. & Dumonceaux J., *Front. Genome Ed.* (2022)

<https://neuromuscular.wustl.edu/pathol/fsh.htm#fshmild>

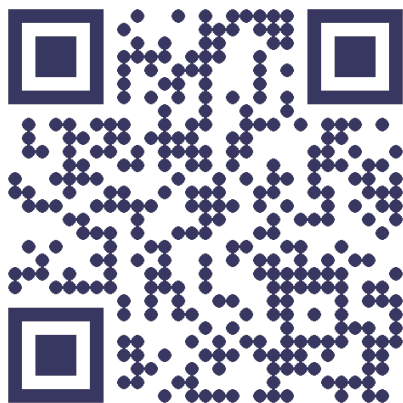
Finding biological mechanisms of FSHD1

What are biological mechanisms of FSHD1 involving
DUX4?



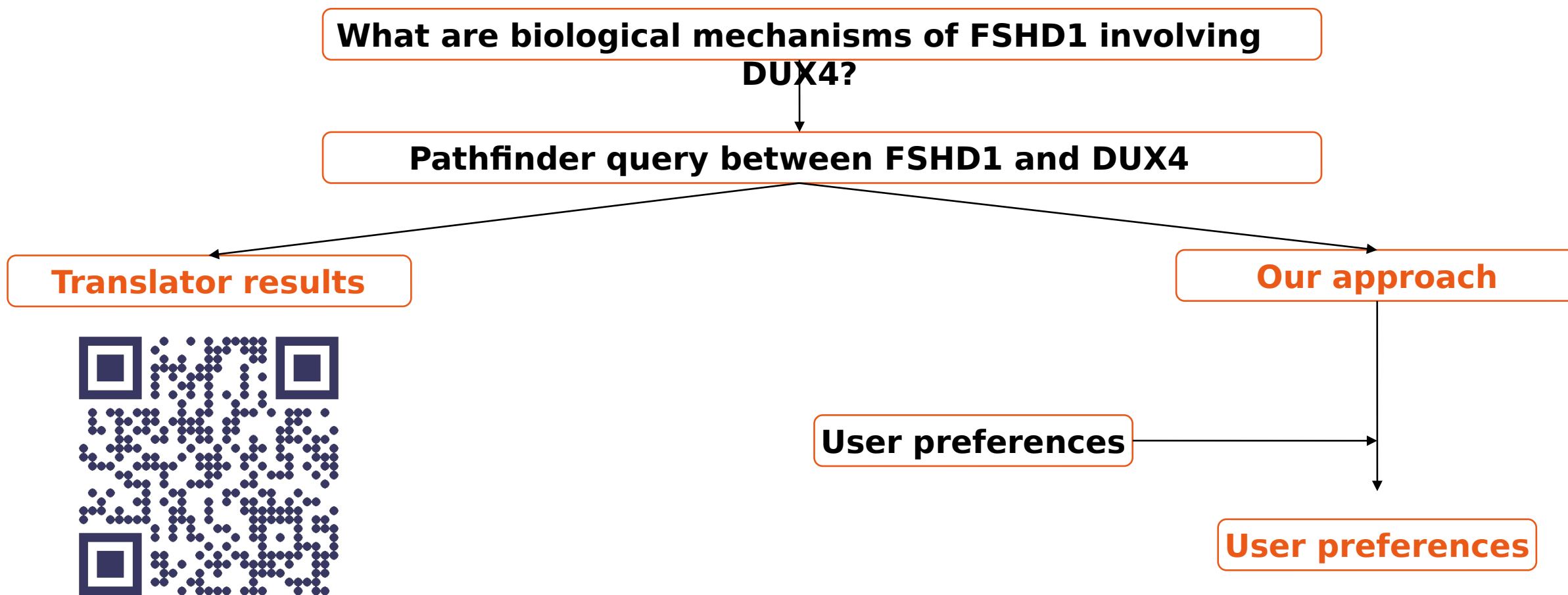
Pathfinder query between FSHD1 and DUX4

Translator results



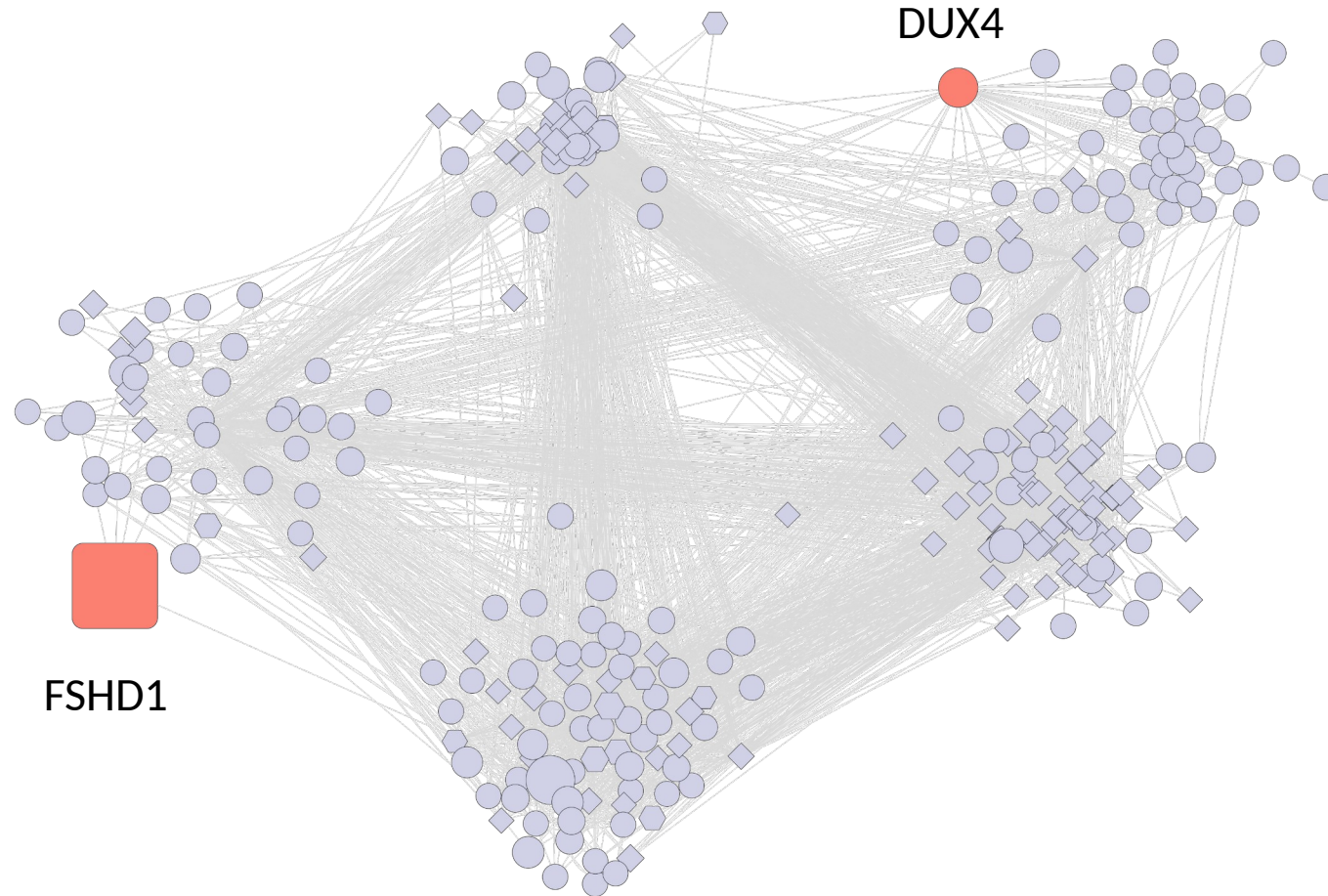
<https://ui.transltr.io/>

Finding biological mechanisms of FSHD1

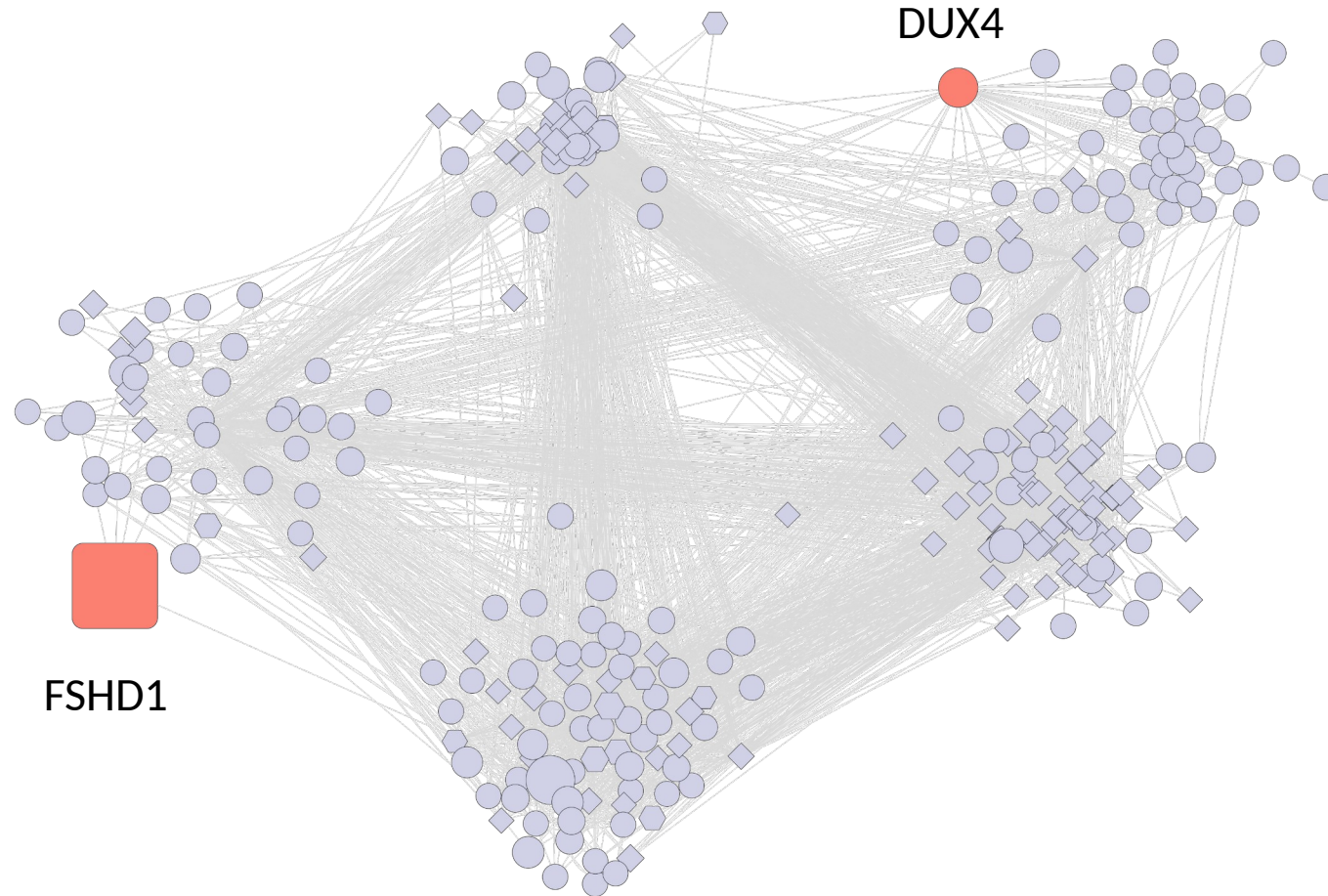


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Finding biological mechanisms of FSHD1

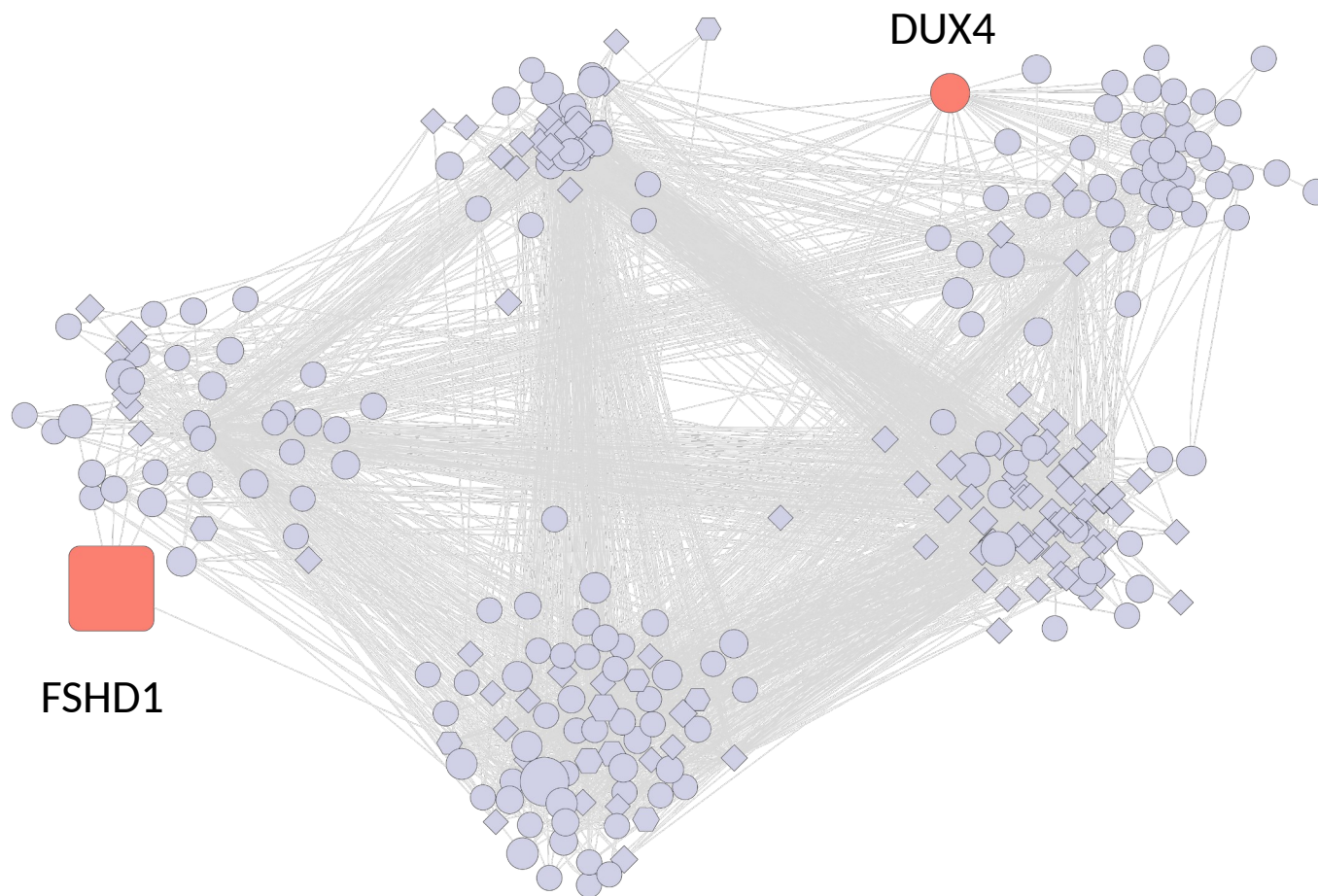


Finding biological mechanisms of FSHD1



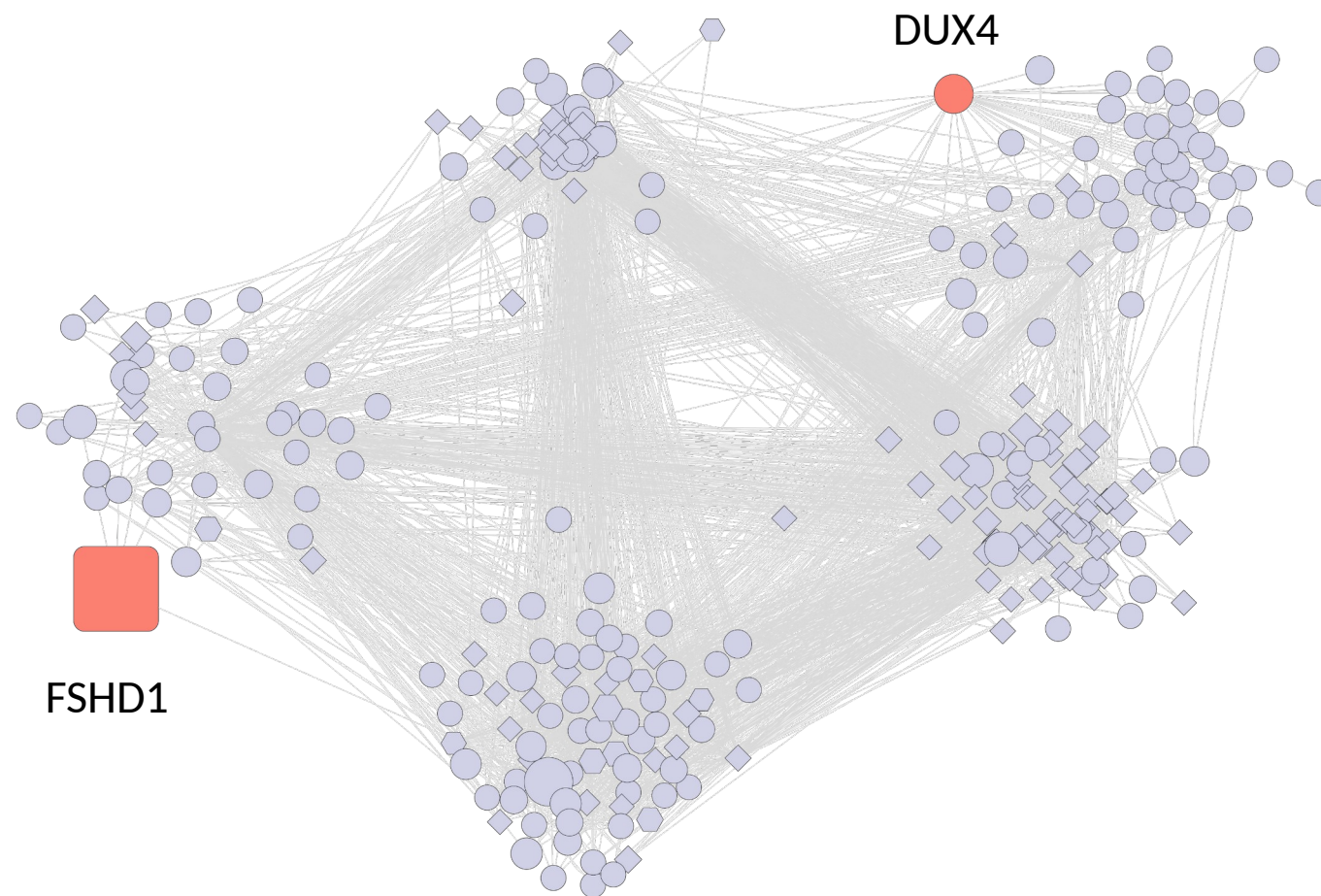
✓ 5 repurposing drugs candidates

A use case: FSHD1 rare disease



- ✓ 5 repurposing drugs candidates
- ✓ 9 putative targets

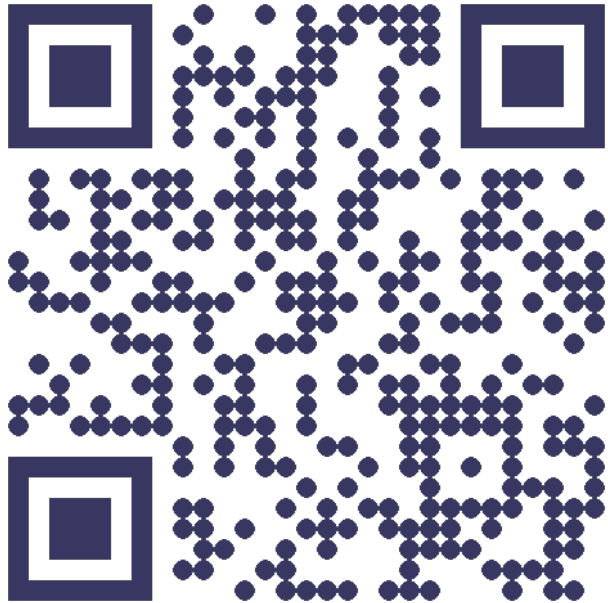
A use case: FSHD1 rare disease



- ✓ 5 repurposing drugs candidates
- ✓ 9 putative targets
- ✓ 1 associated biological mechanism

Accelerating discovery in biomedical sciences

NCATS **Translator**

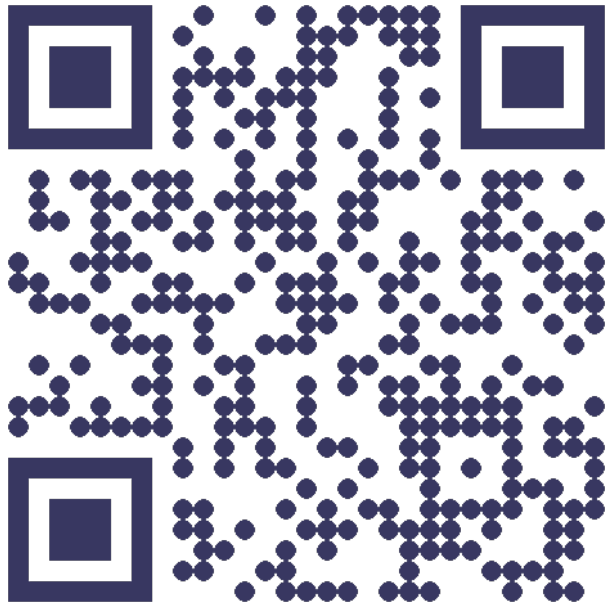


<https://ui.transltr.io/>

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Accelerating discovery in biomedical sciences

NCATS Translator



<https://ui.transltr.io/>

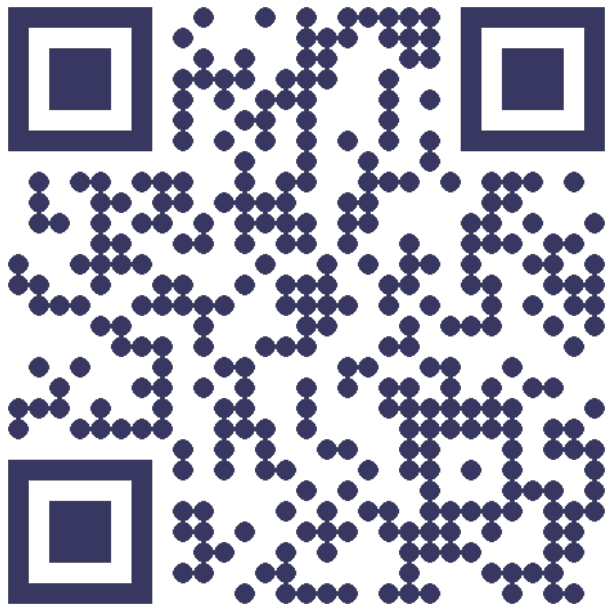
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Accelerating discovery in biomedical sciences

NCATS Translator



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source community!**



<https://ui.transltr.io/>

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sandrine.muller.research@gmail.com