Interoperable Data Sharing in Pediatric Genomics

What is the Gold Standard?

Presented by Vasiliki Rahimzadeh, PhD

STANFORD CENTER FOR BIOMEDICAL ETHICS

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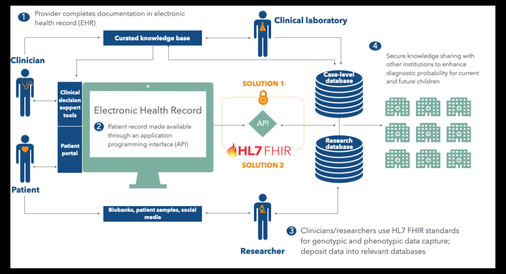
Genetics: Ethical Aspects II

Outline

Part I



Key Implications of Data Sharing (KIDS) framework

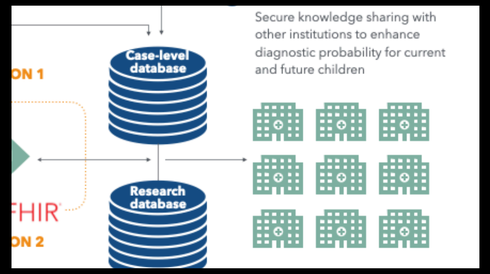
Findings of a policy Delphi study engaging doctors, researchers, ethicists and regulators about responsible data sharing practices in pediatric genomics

Part II

Genomics-enabled learning health system

Ethical challenges and opportunities of sharing genomic & related health data across clinical and research environments

Part III



Work in Progress

Areas for improvement in how genomic data are accessed, used and exchanged at the point of care

GENOMIC AND ASSOCIATED PHENOTYPIC
DATA DRIVE INNOVATIONS
TO PUSH HUMAN HEALTH

More complex genomic and related health datasets are being generated than can be securely accessed and shared, limiting scientist's ability to harness the true power of precision medicine.

Interoperable Data Sharing in Pediatric Genomics: What is the gold standard?

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Genomic data are highly identifiable

Requiring institutional oversight to ensure only authorized users are permitted to access datasets for approved purposes

Children are a protected research population

- Consent to data sharing provided by their parents or other guardians
- Jurisdictional differences in data protection standards
- Limited ability to make difference data sharing choices
- Longer exposure to informational risks

Ethical/Legal

Image credit: Politico

WWW.PROJECTPEDIGREE.ORG

Key Implications of Data Sharing (KIDS) Framework for pediatric genomics

12 policy statements generated from a systematic review of the bioethics literature and following an consensus committee meeting of members of the Global Alliance for Genomics and Health

RAHIMZADEH, V., SCHICKHARDT C, KNOPPERS, ET AL. 2018 *JAMA PEDIATRICS*, *172*(5), PP.476-481.

Perceived likelihood of re-identifying individuals from aggregate datasets, mediated by the openness of the access regime, shape how ethics oversight bodies perceive data sharing risks

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Suboptimal workflows for integrating genomic data in electronic health records impede clinical decision making at the point of care, especially for undiagnosed children

www.projectpedigree.org

RAHIMZADEH, V., BARTLETT, G. AND KNOPPERS, B.M., 2021. *BMC MEDICAL ETHICS*, *22*(1), PP.1-12.

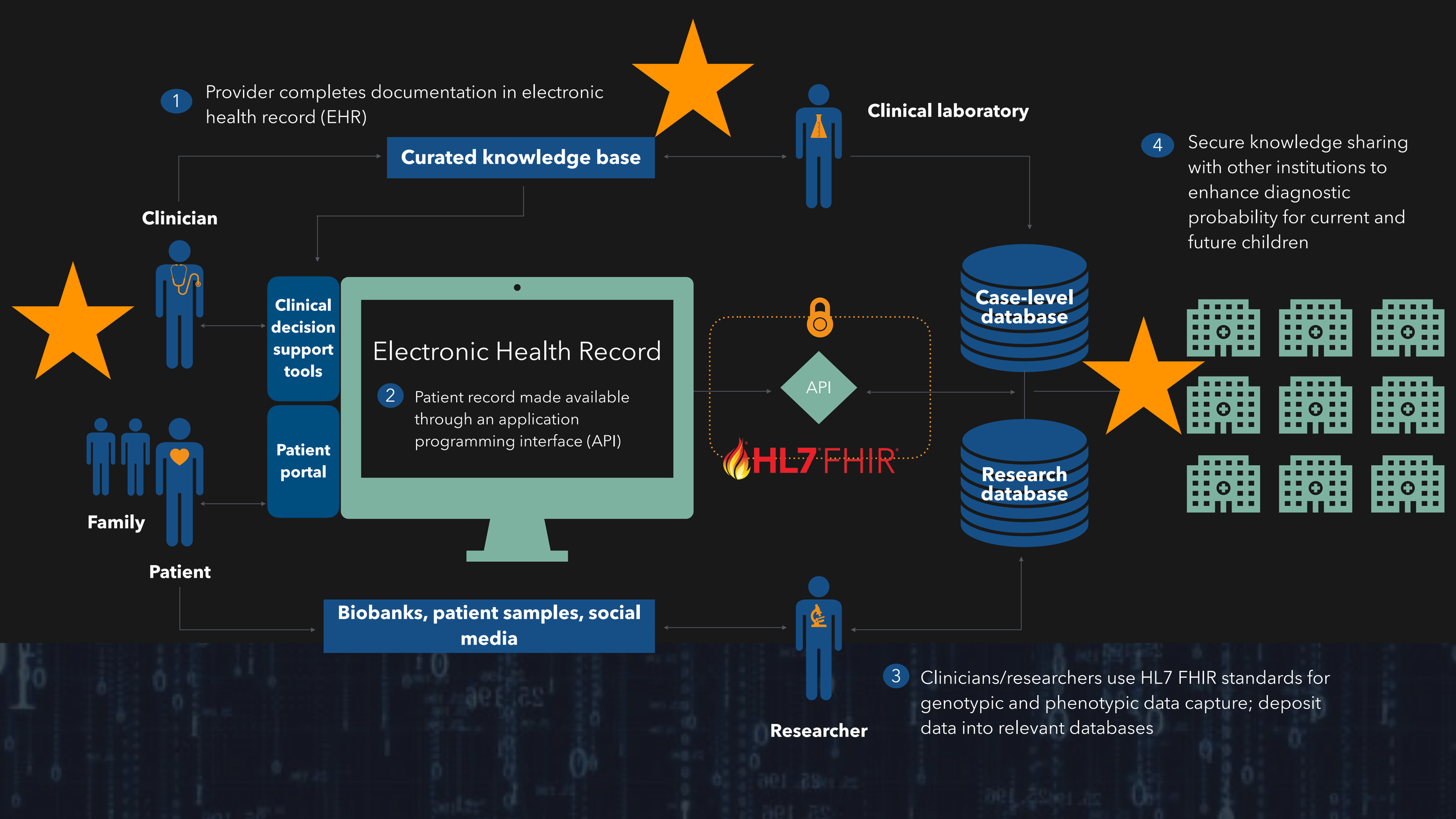
Policy Delphi

3-round policy Delphi study to validate KIDS Framework with an expert panel of Canadian ethicists, medical geneticists, genomic researchers

Anonymized pediatric data should be made available via *publicly accessible* databases

Identifiable pediatric genomic and associated data should be coded and made available through a controlled or registered access process

Providing children and their parents the opportunity to share genomic and associated clinical data is an obligation of those who generate such data


1 Provider completes documentation in electronic health record (EHR)
Curated knowledge base
Clinical laboratory
4 Secure knowledge sharing with other institutions to enhance diagnostic probability for current and future children
Clinician
Clinical decision support tools
Patient portal
Electronic Health Record
2 Patient record made available through an application programming interface (API)
API
HL7 FHIR
Case-level database
Research database
Family
Patient
Biobanks, patient samples, social media
Researcher
3 Clinicians/researchers use HL7 FHIR standards for genotypic and phenotypic data capture; deposit data into relevant databases

unFAIR data

- Developing secure computing platforms for co-locating data access, analysis and sharing
- Data may be **f**indable and **a**ccessibility but rarely **i**nteroperable
- Merging clinical with research datasets

Technical

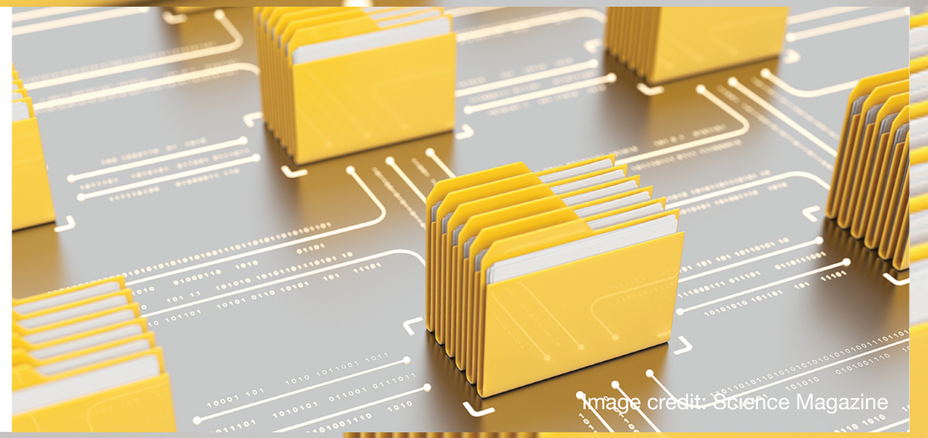


Image credit: Science Magazine

Challenges to genomic data sharing

- Heightened public consciousness to the realities of data privacy, misuse and bias
- Underrepresentation of non-European genomes/diverse genetic ancestry
- Right to an (and private?) future
- Relationally of genetic data and implications for biological relatives

Social.

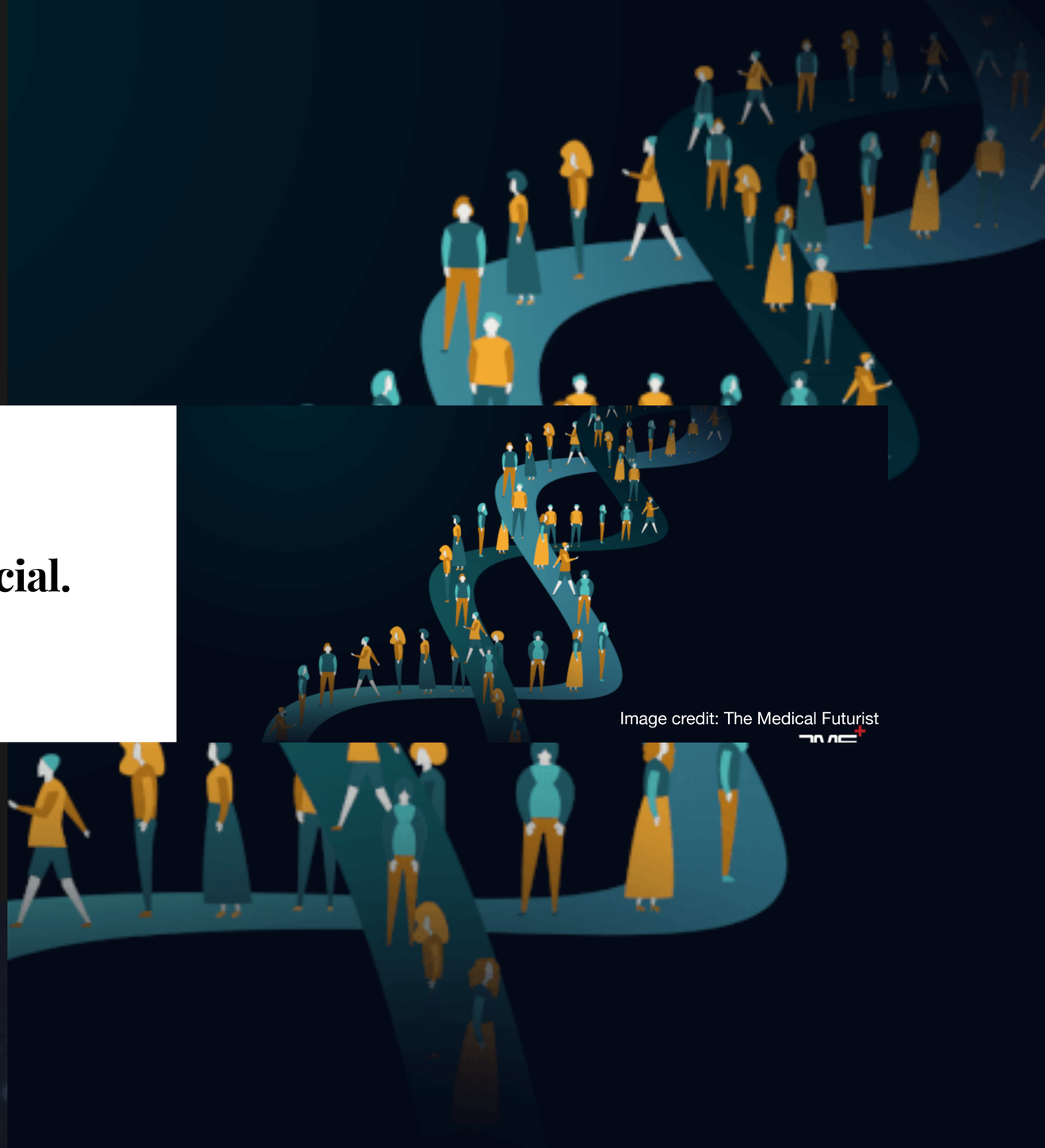


Image credit: The Medical Futurist

How usable are genomic data exchange standards for pediatricians treating undiagnosed children with rare genetic disease?

The benefits of using common standards for data capture and exchange are well recognized, but their implementation in real world, pediatric clinical contexts are significantly understudied despite plans for nation-wide adoption in the coming years.

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Work in progress

A random sample of 20 ≤ pediatric medical geneticists and developmental pediatricians will participate in an online simulation of a prototypical EHR workflow using the ClinGen's HL7 FHIR® -compliant genomic resources.

PARTICIPANTS WILL 'SOLVE' A GENETIC DIAGNOSIS INVOLVING A PEDIATRIC PATIENT AS PART OF A CLINICAL VIGNETTE

USABILITY METRICS DRAWN AT FUNCTIONAL POINTS IN THE EHR WORKFLOW

- **SITE NAVIGATION**
- **DATA UPLOAD**
- **DATA RETRIEVAL**
- **CLINICAL NOTATION**

SYSTEM USABILITY SURVEY ON USER EXPERIENCE

Expected Outcomes

- Identify the least/most usable features of existing data exchange standards among practicing clinicians
- Make targeted recommendations to guide developers on usable design specific to pediatric use cases

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Thank You.

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