



Considerations for developing a national genomic data base and sharing of data in Brazil

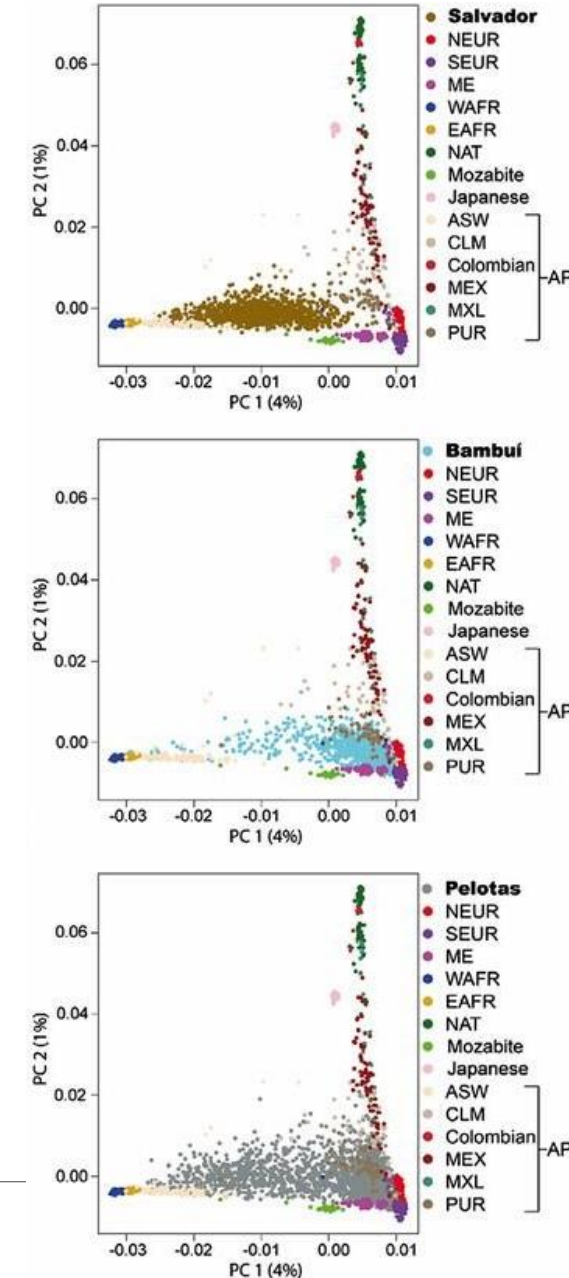
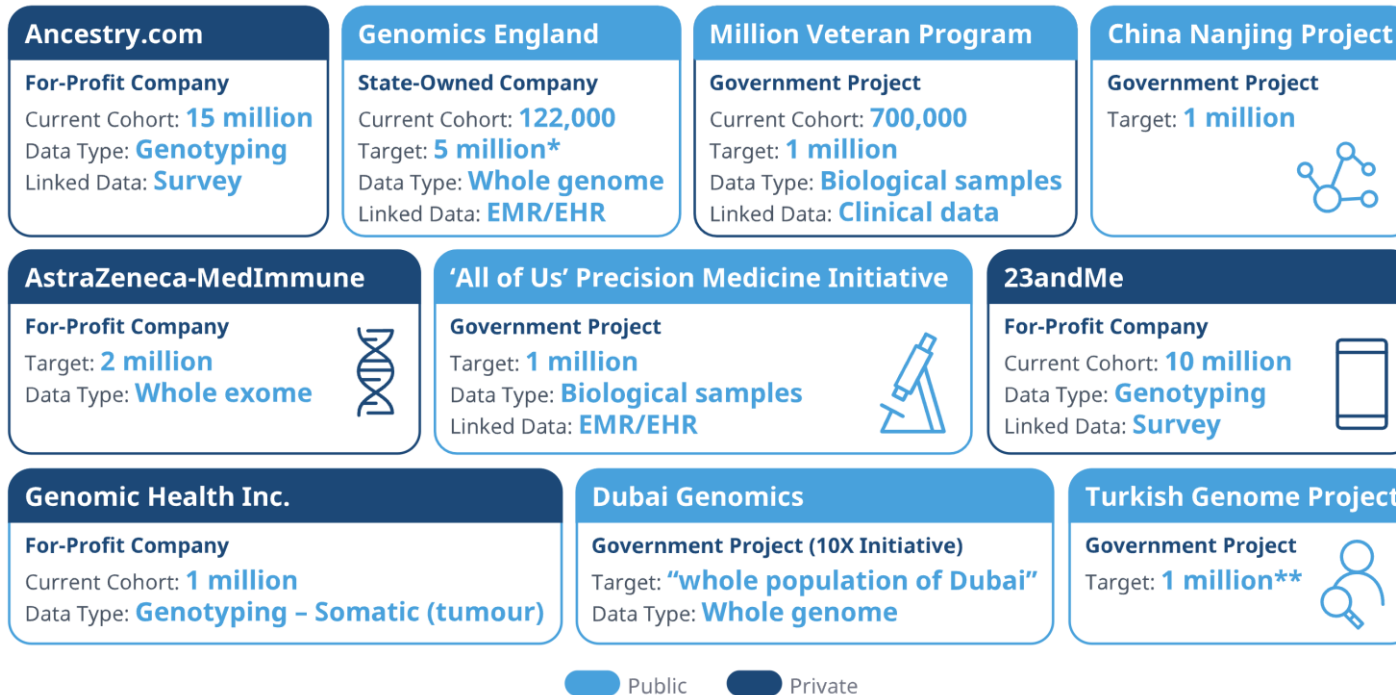
Pablo Ivan Pereira Ramos

16th May 2024

Regional meeting
**Human Genomics for Health:
Enhancing the Impact of Effective Research**

Contextualization

- **Brazilian population**—marked genetic diversity resulting from the country's historical construction and migration patterns, added to ~5 centuries of admixture¹.
- **Many international experiences in human genomics², e.g.:**



¹ Kehdy et al. PNAS. 112 (28) 8696-8701, 2015.

² IQVIA Genomic initiatives database

Contextualization

- **Genomas Brasil**—will sequence 100,000 individuals, with focus on different diseases (rare/monogenic, oncological, cardiometabolic, infectious, and others) as well as populational samples.
 - Lay groundwork for future incorporation of precision health into Brazil's Unified Health System (SUS)
 - Accelerate the development of the genomics national industry



Contextualization

- **Brazil's specificities**
 - **Regulatory:** The LGPD¹ considers genetic data as sensitive personal information.
 - **Ethical:** Nat'l. Health Council's (CNS) Res. 738/2024 & other infralegal normatives.
 - **Cultural:** genomic studies on an ethnically diverse population (e.g., indigenous, *quilombolas*) should be conducted in a culturally-sensitive manner.
 - **Health system configuration:** architectural complexities of SUS and where to plug precision medicine into clinical practice—RNDS as potential interoperability bus.

¹ Law 13.709/2018

Towards GenBRdb Biobank

- **What are our requirements & priorities?**

- Researchers & genomics community
- Clinicians
- Society

Workshop held in
Brasilia, October 2023

- **Different architectural and infrastructural options**

- On-premise
- All-cloud
- Hybrid

Mining of papers/gray literature, interactions
with other nat'l initiatives

- **No *one-size-fits-all* approach suffice,
but specialized platforms can be used to
accelerate data sharing and re-use**

Interactions and demonstrations with vendors of
specialized analytical platforms

Towards GenBRdb Biobank

- **3-day summit in Brasilia**—discussion of key needs and requirements to subsidize the construction of the Analytical Platform and Data Repository of the *Genomas Brasil* program

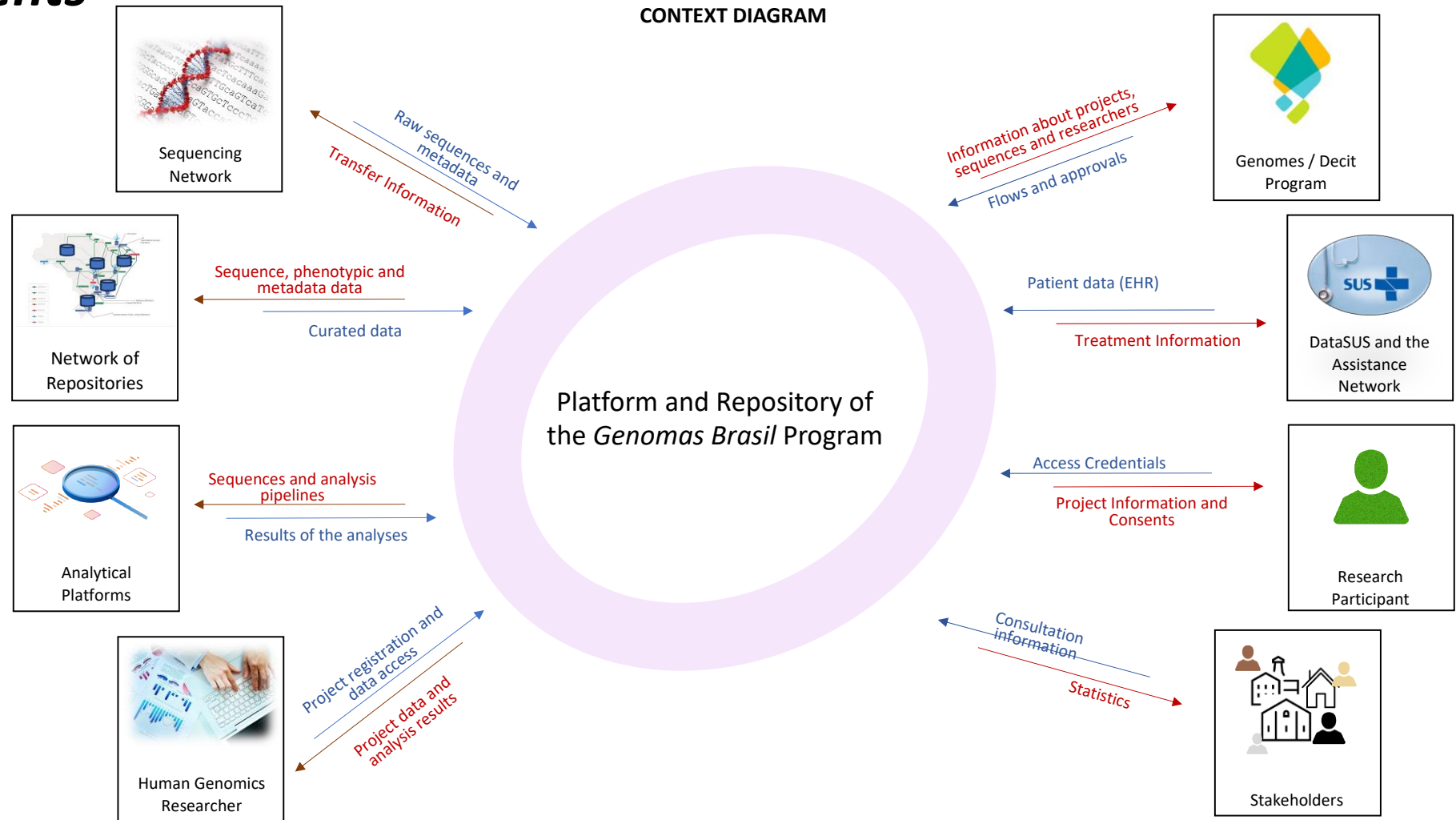
Requirements

- Standards to enable **interoperability**, e.g. GA4GH
- Management of the **informed consent** (e.g. through dynamic consent)
- Establishment of the **data governance** for the initiative
- Linkage** of the genomic data with other administrative/health databases
- Virtual cohorts** to accelerate data re-use
- Data **redundancy** to safeguard this strategic asset
- Access** passports to verified researchers
- A **hybrid storage model**, implemented in a phased way
 1. On-prem guarantees sovereignty and security
 2. Public cloud guarantees scalability and sharing of (less sensitive) data



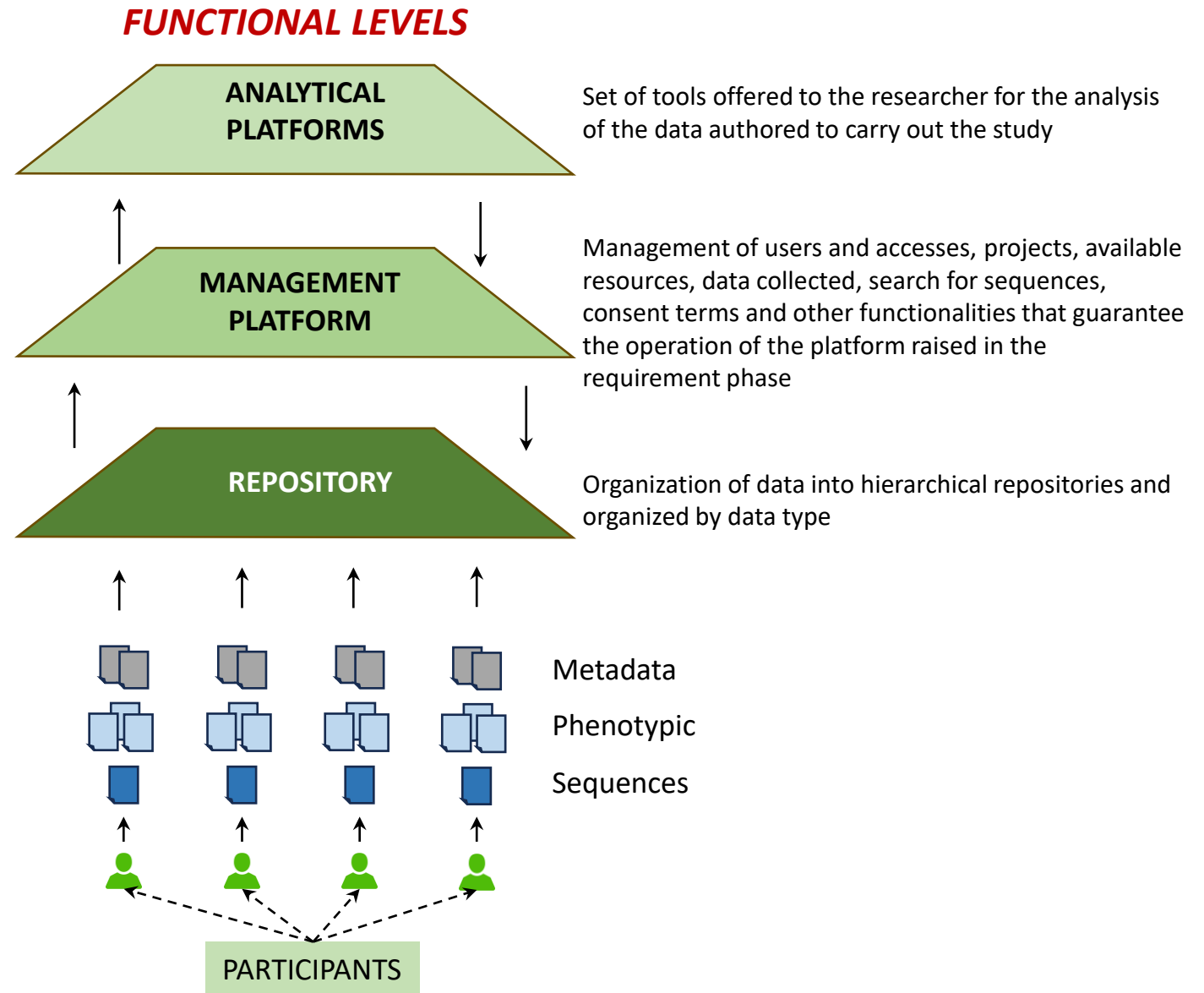
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- **The components**



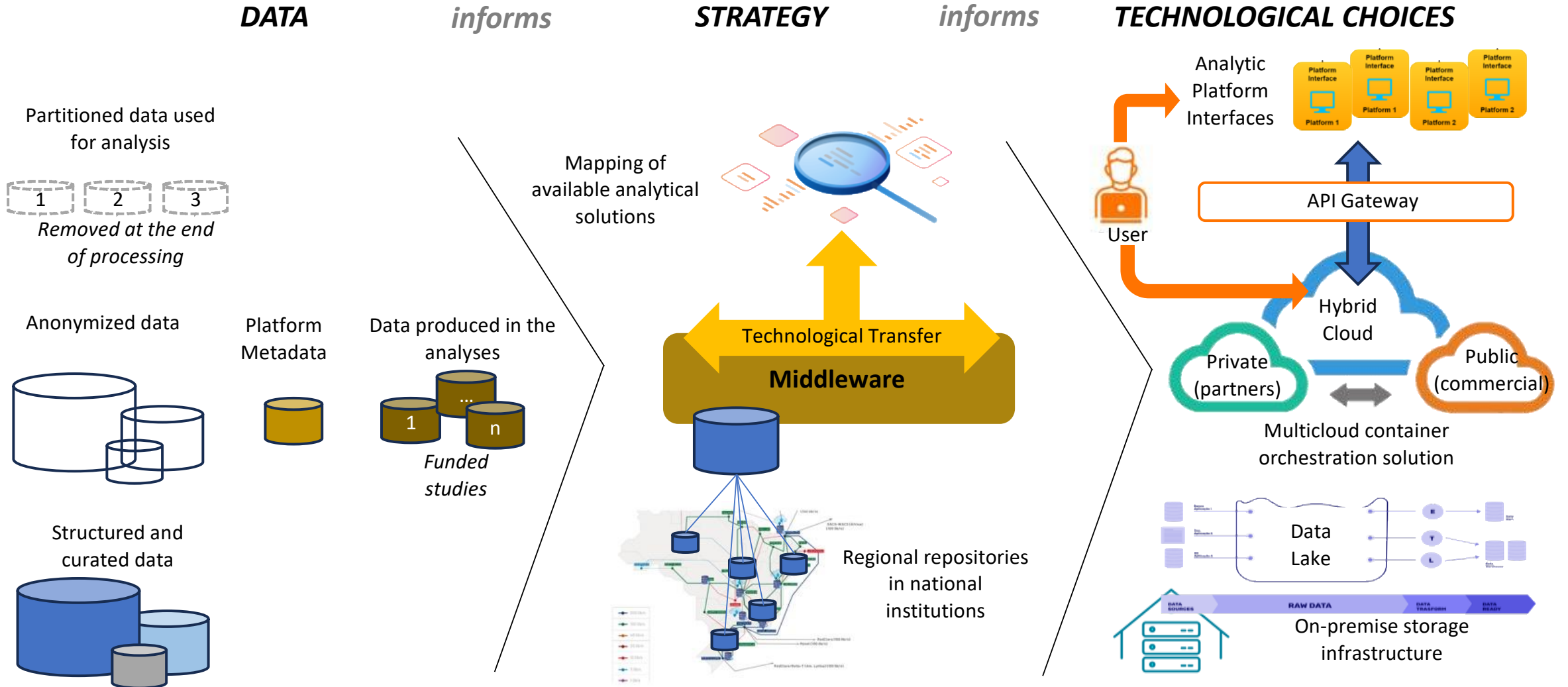
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- The *architecture* (3-tiered)



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- The *vision*



Challenges Ahead

- **Sharing, reuse, ethics**—how to balance the need to put nat'l scale genomic data to use (informing therapy; diagnostics; new research questions) in an ethically-sound manner and abiding to Brazil's regulatory environment.
- **Data governance**—need to establish flows, authorized users, and approval instances.
- **Nat'l. data infrastructures**—leverage existing networks and investments in this area.
- **Data sovereignty and security.**
- **From *competition* to *coalition***—cultural changes to data sharing, considering this strategic data asset.



Obrigado

Gracias
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