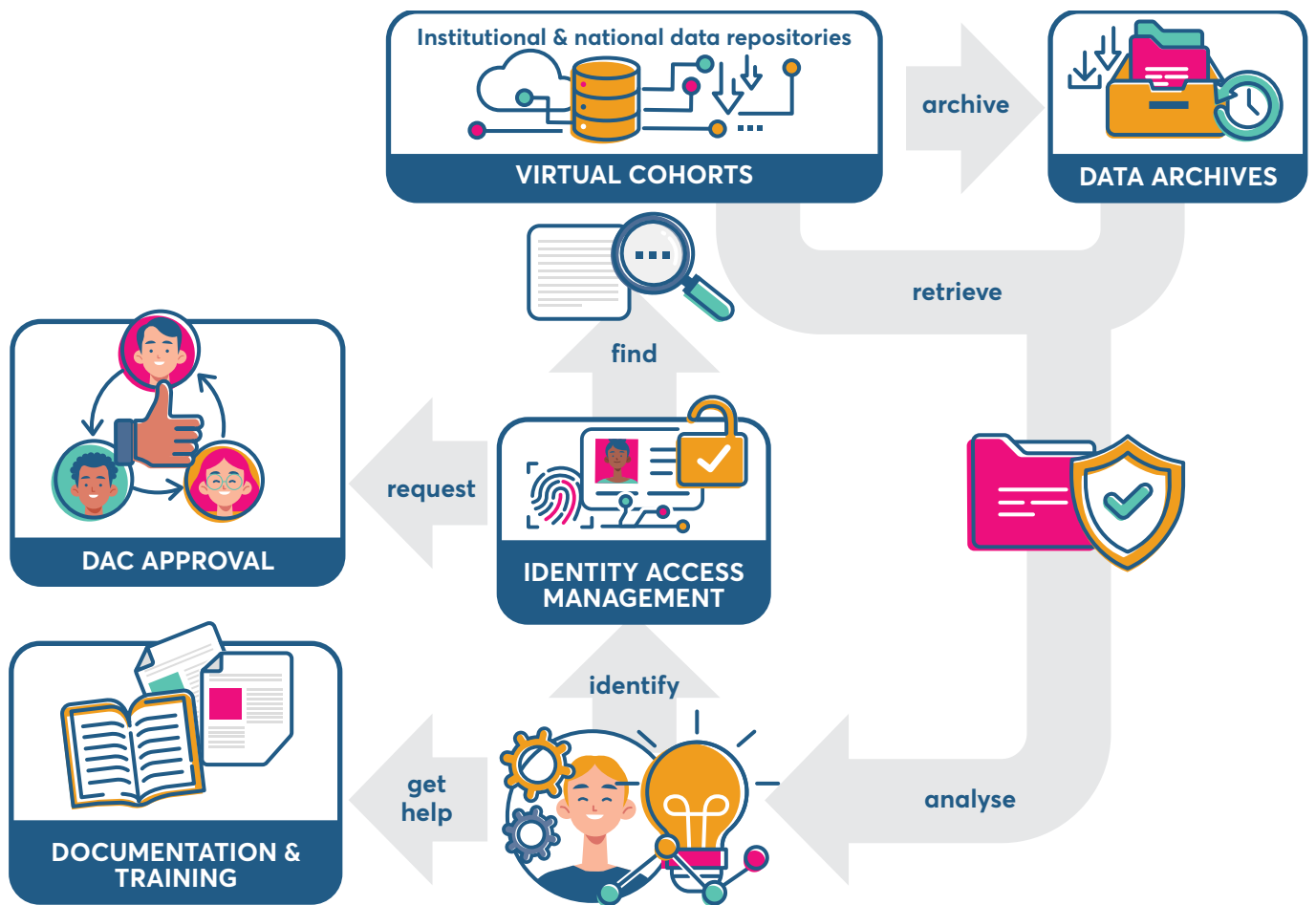# Australian BioCommons

Human Genomes Platform Project

Australian BioCommons is establishing infrastructure for human genome data warehousing, sharing and analysis in Australia that adheres to various global best practice standards.

Working with key research partners, respected infrastructure providers and expert international groups establishing aligned systems, the **Human Genomes Platform Project** is building the necessary foundations for Australia's full participation in the global ecosystem of responsible human genomics data sharing and analysis.

Key challenges have been investigated and candidate solutions tested in pilot implementations. In assessing options, core business requirements of partner institutes were explored to ensure the leading system could be implemented across organisations.

DISCOVERING VIRTUAL COHORTS

Australian human genomic data is siloed and there is no national solution for Australian researchers to identify cohorts of individuals who have had their genomes sequenced.

The identification of cohorts of individuals across multiple repositories enables researchers to discover data to integrate, analyse and derive new insights.

Beacon v2 is a GA4GH standard that has been adopted by major projects globally.

Beacon provides:

- a good combination of clinical/demographic detail and genomic information
- both a framework for the API, as well as a common data model
- flexibility in access control restrictions
- deployment as an individual Beacon, or as a network.

STREAMLINING THE DATA ACCESS COMMITTEE PROCESS

The Data Access Request and Approval process undertaken by Data Access Committees (DACs) requires significant human effort, making it time-consuming and burdensome.

Streamlining the process driven by the DACs will expedite requests and approvals and enable faster, safer and more auditable data access for Australian researchers.

The open source and actively maintained Resource Entitlement Management System (REMS) software is part of the ELIXIR Compute Platform and core infrastructure for major European projects such as the Genome Data Infrastructure.

REMS provides:

- single sign-on integration for increased trust and reduced need for separate user IDs
- configuration with custom forms and workflows to meet the needs of the DAC and a dataset's Data Use Policy
- a comprehensive REST API to allow for integration with other software and automated reporting
- deployment capabilities on a variety of compute infrastructures within Australia.

IMPROVING HUMAN GENOMICS DATA & METADATA ARCHIVING

The majority of Australian human genomics research data is archived in international databases like the European Genome-phenome Archive (EGA). The distance travelled by this sensitive data creates upload and download speed limitations and potential legal issues.

A platform based on the federated EGA model transfers metadata between the central and federated nodes, enables global accession assignment, standardised metadata and enhanced discovery, without sensitive human data leaving the country. A national controlled access data repository for human genomics data in Australia could become one of more than 20 countries adopting this mature and growing model.

A national human genomics repository working within the federated EGA model would:

- be deployable locally with full control over platform and data hosting
- offer existing guidelines, policies, standards, templates and advice to build upon
- have longevity given it underlies national infrastructure in several countries
- provide a solid framework to integrate with future national scale human 'omics infrastructure.

FEDERATING IDENTITY AND DATA ACCESS MANAGEMENT

It is critical to identify and trust users before giving them access to sensitive human genomics research data.

Establishing a high level of trust across services by leveraging existing trusted institutional identities diminishes the burden of establishing identity and eliminates the need for multiple identities.

The CILogon platform allows for identity and access management, providing federated identity management as well as specific group management.

CILogon provides:

- validation of identity, professional credentials and assigned role
- compatibility to a wide variety of stakeholders requirements.

CILogon has been deployed within Australia and managed through project partner, Australian Access Federation (AAF).

Australian BioCommons and the HGPP partners can help you:

- deploy your own Beacon, submit metadata to an existing Beacon or add an existing Beacon into the network
- deploy your own REMS instance, manage your existing DAC with REMS, try out a demo REMS instance
- start using CILogon for authentication for your service, investigate other federated identity solutions, or connect your institute to the AAF.
- advocate for what you would like to see in a national human genomics repository

Findings, recommendations and detailed documentation are available to everyone.

Learn more about the Human Genomes Platform Project by visiting **biocommons.org.au** or reach out to us: **contact@biocommons.org.au**. We'd love to hear from you!

AUSTRALIAN ACCESS FEDERATION

Australian Genomics

ARDC Australian Research Data Commons

BIOPLATFORMS AUSTRALIA

Children's Cancer Institute | ZERO CHILDHOOD CANCER | KIDS CANCER CENTRE SYDNEY CHILDREN'S HOSPITAL

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