The American Journal of Human Genetics, Volume *105*

Supplemental Data

Australian Genomics: A Federated Model for Integrating Genomics into Healthcare

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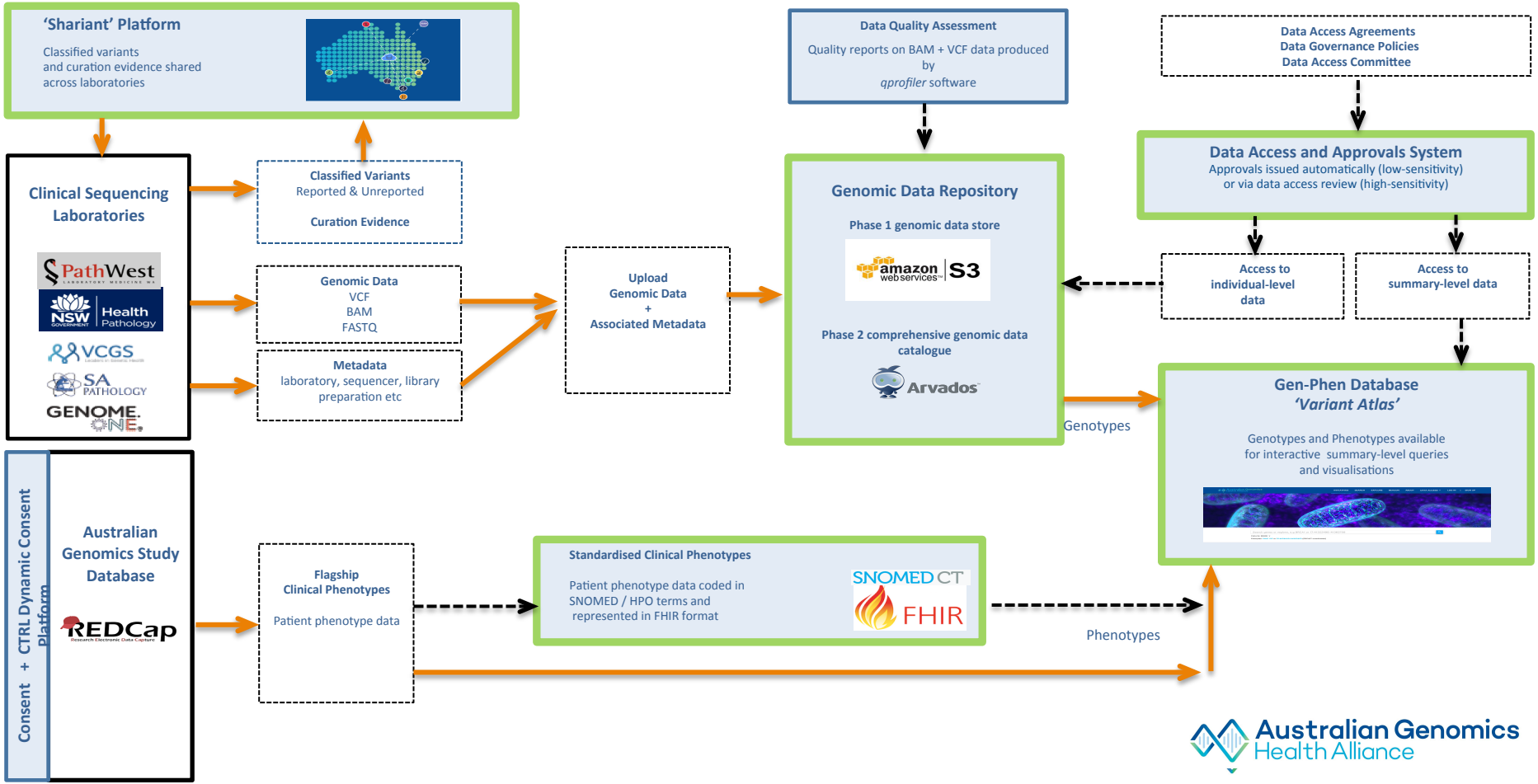
Figure S1: Australian Genomics data management work flow and infrastructure



Supplementary Table 1: *Australian Genomics* programs and sub-projects.

PROGRAM 1: National diagnostic and research network	PROGRAM 2: National approach to data federation and analysis	PROGRAM 3: Evaluation, Policy and Ethics	PROGRAM 4: Workforce and Education
National clinical genomic consent Develop a single shared consent process and supporting documentation for clinical genomic testing for all indications.	**Study database (REDCap)** Manage participant clinical, demographic and survey data	**Health implementation research** Evaluate progress across the Flagship projects to ensure research outcomes are being effectively translated into practice. Create a research base for getting evidence into practice.	**Workforce status** Professional status survey of genetic counsellors and clinical geneticists, including current activity and education in genomics.
CTRL – Participant portal and dynamic consent platform Empower research participants with granular, dynamic consent options; updates about the research they've contributed to; and access to information, and patient support. Real-time feed-back loop to researchers as consent preferences change.	**Standardized, computer-readable clinical data** Translate clinical data into standardized codes (phenotype ontologies: HPO/SNOMED) to allow computerized search, discovery and coding	**Social network analysis** Map and analyze interconnections between members to capture the collaborations amongst the genomic community, to document learning, assess Australian Genomics' influence, and identify key players in implementation.	**Existing genomics education** Map existing Australian education and training for health care professionals.
Virtual gene panels Develop a national approach to	**Genomic data repository** The cloud-based storage of	**Genomics policy** Mapping of major genomic	**Needs assessment** A portfolio of projects aiming to understand health professionals'

the design, maintenance and use of virtual gene panels in genomic analysis to minimise duplication of effort and ensure consistency of data analysis.	genomic sequencing data (BAM/VCF/FASTQ) together with a subset of the participant's clinical data to facilitate meaningful analysis and sharing.	medicine policy frameworks and demonstration projects internationally. Review of the international genomics policy landscape, including reimbursement policies, genetic discrimination and data sharing.	current practice and needs for continuing medical education in genomics, including genetic specialists, medical specialists, and community practitioners from the perspectives of these professionals, educators and patients.
Clinical variant re-classification Develop guidelines for managing re-classification of genetic variants, and systems to ensure reporting to referring clinicians and patients	**Genotype-Phenotype platform: Variant Atlas** Interactive Genotype-Phenotype data platform to visualise aggregated variant data, and filter by key clinical features to describe cohorts	**Ethics and legal** Review and make recommendations on the legal and ethical aspects of various models and applications of genomic data sharing in Australia and internationally Contribute to Australian Genomics initiatives in clinical consent, research consent and insurance discrimination.	**Evaluating Education** Develop and pilot tools to support development of effective genomic education strategies and enable evidence-based education practice.
Mainstreaming genomic pathology reports Develop recommendations for plain-language genomic test reporting, making reports accessible to non-genetic health	**National variant sharing platform: Shariant** Online platform for laboratories to share curated variant classifications and detailed curation evidence in real-time	**Health economic evaluation** Measuring and valuing the short- and long-term benefits of clinical genomics using diverse approaches including cost-effectiveness, cost-benefit, cost-	**Genetic Counseling for rapid turnaround.** Examine genetic counselling processes in the acute care setting and the psychosocial impact of rapid genomic

professionals, and patients	i.e. prior to report generation. Automatic notification about classification differences, and submission of summary information to international databases.	utility, and budget analyses.	sequencing on families to inform service delivery models.
MSAC application pipeline Apply evidence built through clinical Flagships to submit applications to MSAC seeking Federal funding recommendations for genomic testing	**Data access agreements and policies** Participants provide consent to both national and international data sharing, for the benefit of healthcare. Clinicians and researchers can request access to Australian Genomics datasets, and this will be granted according to the level of data sensitivity, the specific consent of the participant, and the researcher's HREC, where applicable. Australian Genomics subscribes to GA4GH policies and standards.		**Public perceptions and expectations** Establish an evidence base to understand the Australian public's diversity of experiences, values, attitudes and expectations of genomics in research and healthcare.
Functional genomics network Linking clinicians and researchers, this project aims to provide catalyst grants for pilot			

projects aiming to incorporate functional genomic assays into the diagnostic pipeline			
Genomics in the community Develop a suite of information resources about genomics, genomic testing and related issues (eg data sharing), in collaboration with patient advocacy groups			