GENYSIS: a novel core facility for clinical evaluation of research genomic sequence

Bradford Powell[1], Kimberly Foss[1], Elizabeth Hutchins[1], Scott Melville[1], Mai Xiong[2], Tam Sneddon[2]

[1]Department of Genetics, [2]Department of Pathology and Laboratory Medicine; University of North Carolina at Chapel Hill School of Medicine

Introduction

- Over multiple research studies, our group has developed expertise, infrastructure and protocols, and reporting of clinically-relevant variants from research-derived genetic data

NCGenes NCNexus North Carolina Newborn Exome Sequencing for Universal Screening

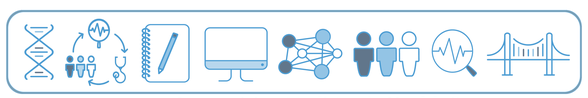
- Genetic data are increasingly incorporated into research studies by investigators whose primary expertise is outside the domain of clinical variant classification
- The **Clinical GENomic AnalYSIS** (**GENYSIS**) core facility was formed to disseminate the ability to incorporate analysis and return of these results to such studies

Clinical Genomic Analysis as a Research Service

- GENYSIS uses a "research recharge center" model
- The fee-for-use model enables investigators to budget based on number of participants and expected yield of returnable findings, rather than budgeting for effort for key individuals
- We encourage study investigators to participate in the multidisciplinary conference where their cases are discussed
- Initial service rates were based on costs from historical studies and anticipated demand- rates will be reviewed annually
- Diagnostic and/or secondary findings may be returned to research participants and/or the health record, depending on study design and participant consent
- We envision that this model could be useful for development of similar services in other institutions

Ongoing Research and Development Projects

- Evaluation of study protocols and consent forms that include return of clinically-reportable results of genome-scale sequencing. We plan to produce recommended language to include as part of study documents
- Incorporation of short-read and long-read-based techniques to detect and classify copy-number and structural variants
- Development of techniques for investigators to communicate structured information about participant phenotype to incorporate in our analyses



GENYSIS is supported by the University of North Carolina Departments of Genetics and of Pathology and Laboratory Medicine, and from the Program for Precision Medicine in Health Care

med.unc.edu/genysis

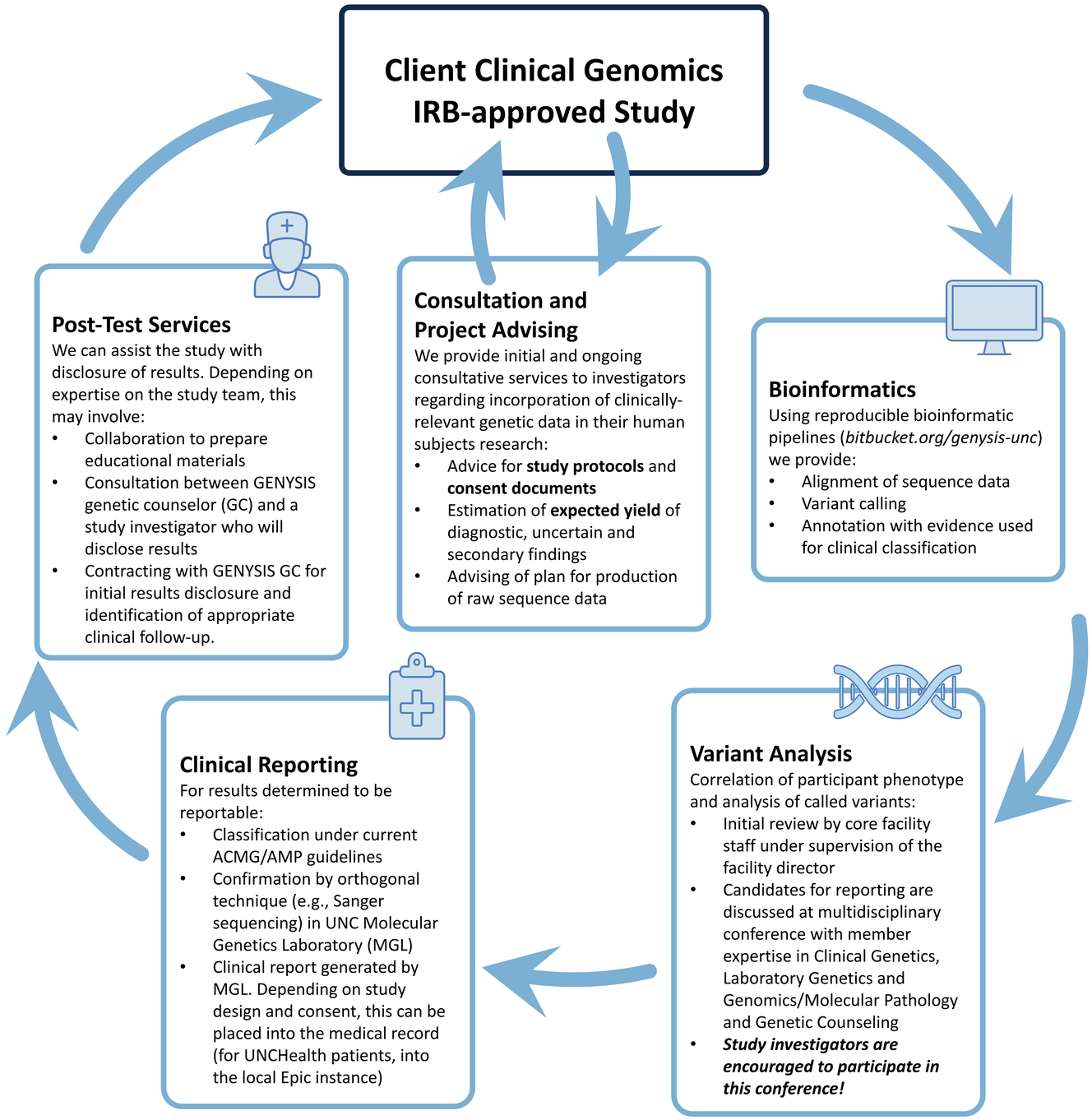


Figure: Workflow in the GENYSIS core facility. All analysis takes place for research participants in studies with appropriate consent and approval by Institutional Review Board (IRB). We focus on the analysis of these data for clinical relevance and do not duplicate services provided by other core facilities at our institution: the Biospecimen Processing (**BSP**) and High Throughput Sequencing Facility (**HTSF**) can be used for DNA extraction and production of primary sequencing data, respectively. Alternatively, investigators can provide sequence data or variant calls from their own analysis pipelines for analysis and classification. Our core services are described in light-blue boxes.