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Precision Public Health in Action: New CDC Pilot Projects Integrate Human Genomics into Public Health Surveillance and Applied Research

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In collaboration with the CDC Office of Advanced Molecular Detection, we recently launched a new, five-year initiative to strengthen public health capacity by introducing elements of human genomics into both public health surveillance and applied research. We report here on the successful launch of one of the initiative's components.

The Office of Genomics and Precision Public Health (OGPPH) put out a call for pilot exemplar projects throughout CDC and ATSDR Centers, Institutes, and Offices (CIOs). The process yielded 23 applications from across CDC, covering infectious and noninfectious disease topics throughout the lifespan.

Applications were reviewed by OGPPH staff and an objective review panel. The reviewers ranked the proposals according to six criteria: scientific merit, alignment with public health genomics priority areas, potential public health impact, focus on health equity, potential for growing program capacity, and feasibility.

Six projects across five CIOs were selected for funding in 2022 and 2023. They cover a wide range of topics, including:

- Assessing the impact of genetics in the control of two infectious diseases (Tuberculosis and Ebola),
- Enhancing the reporting of gene/genome sequencing in newborn screening programs,
- Examining the role of medications and genetics in the National Birth Defects Prevention Study (NBDPS),
- Establishing population-based, ethnicity-specific allele frequencies for pharmacogenomic traits of public health importance using the National Health and Nutrition Examination Survey (NHANES),
- Enhancing the evaluation of genetic risk prediction models for inhibitor development among people with hemophilia in different populations.

A summary of these projects can be found in Table 1 below. In the coming weeks, investigators from each project will describe their study and its potential impact.

We are excited to announce these six projects and will regularly report progress in these and other efforts to integrate human genomics in public health surveillance and applied research.

This is just the beginning. We will continue working with the public health community at CDC to strengthen and sponsor other proposals. Our goal is for this process to lead to a new community of practice in genomics and precision public health.

We are interested in your feedback about priority areas for human genomics and public health. Please submit your input below.

Table 1: Six Pilot Projects in Human Genomics and Public Health Surveillance and Applied Research in 2022 and 2023.

Title	Project Summary
Enhancing hemophilia inhibitor risk prediction using genetic risk prediction models in a diverse patient population	Leveraging existing technology and stored specimens, HLA genes and variants of immune response genes (IL-10, TNFA, IL1A, IL12B, and CD80) are to be sequenced and genotyped, respectively, establishing a broader set of Hemophilia Inhibitor Research Study (HIRS) enrollees to evaluate existing inhibitor risk prediction models and to investigate the feasibility of developing better models using curated variants in a diverse patient population.
Impact of pharmacogenetics on tuberculosis drug pharmacokinetics, drug response, and safety in TBTC/ACTG study S31/A5349*	The contribution of pharmacogenetics (PG) will be evaluated in a large population subset from a phase III clinical trial with respect to patient efficacy and safety outcomes by incorporating PG data into population pharmacokinetic (PK) models for six major TB drugs and risk algorithms for unfavorable treatment outcomes and to develop and evaluate PG-based dosing algorithms for novel high-dose rifapentine-based regimens.
Expanding Bioinformatics in Newborn Screening: Increasing the Utility of ED3N**	The enhancement in detection of rare, treatable diseases in newborns will be accomplished through 1) the development of requirements to meet NBS program needs in OAMD's existing LIMS Lite application for connecting public health laboratory sequencing data with CDC, 2) NBS-specific bioinformatics pipeline/s construction, modifications, and validation and connection to the existing ED3N platform for transfer of variant calls and variant interpretation within ED3N. These enhancements will be tested with at least two state NBS programs.
Genetic variation in natural serum immunity against Ebola virus	Mechanisms of unspecific Ebola virus inhibition in certain sera from naïve donors will be determined with exploration of therapeutic potential from results. Human genomic research techniques will be incorporated into existing virology research to determine susceptibility or resistance to Ebola infection.
Pharmacogenetic Allele Frequencies in a Large Population-Based Sample from the United States	The genotype of 150 pharmacogenetic (PGx) genes will be determined for approximately 5,000 NHANES DNA samples and correlated with NHANES data regarding ethnicity and prescription drug usage. These data will help to establish the PGx allele frequency for African Americans, Asians, and Hispanics in the US. The appropriateness of prescribed drugs for will be evaluated and ethnic-specific risk reduction regarding inaccurate drug selection will be assessed with respect to PGx genotype.
Gene Variants and Potential Interaction Effects with Medications and Non-syndromic Birth Defects	Genome-wide genotyping of DNA from NBDPS family trios with maternal periconceptional exposure to antibiotics or antidepressants will be performed. An assay with enhanced pharmacogenomic content will be used to establish drug metabolizing status. Data will be shared with grantees and collaborators for analyses such as GWAS, Mendelian randomization, and polygenic risk score to inform birth defect risk.

*Rifapentine-containing Treatment Shortening Regimens for Pulmonary Tuberculosis: A Randomized, Open-label, Controlled Phase 3 Clinical Trial. TBTC Study 31, ACTG Study A5349

**Enhancing Data-driven Disease Detection in Newborns

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