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Genomic correlations to childhood health outcomes: Integration of clinical and genomic data

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Purpose: "The First Thousand Days of Life" longitudinal birth cohort study provides the largest data bank amassed from clinical and genomic data thus far and will be a cornerstone of pediatric observational epidemiology for many years. This study gathers biological, clinical and social data to assess the development of the mother-child dyad from pregnancy through early childhood. Early childhood health outcomes are shaped by environmental, social, behavioral, genetic and epigenetic determinants. Cost has largely precluded extensive genetic/genomic and epigenetic analyses from large longitudinal cohort studies. However, recent technological advances have made genomic sequencing more affordable and available. We present the methodology, infrastructure and preliminary results following the first 3 years of the largest longitudinal genome-based birth cohort study.

Methods: The cohort goal is 5,000 parent-infant triads. Participating families are enrolled in the 2nd trimester within the obstetric clinics of a single, large healthcare system as well as by use of several forms of geographically and demographically targeted social media. In addition to electronic medical records, we collect parental enrollment questionnaires consisting of self-reported medical history, exposures and lifestyle information, all of which is captured and stored in a custom electronic data capture system using the platform. Additional surveys are collected every 6 months after delivery through a web-based system with email notifications enabling access to a web-based account (paper versions are available for participants without email or web access). Both platforms are integrated into a single clinical trial management system. Biological sample analysis includes triad-based whole-genome sequencing and related RNA/protein/epigenetic analyses. All data storage and analysis is cloud-based for heightened security and patient confidentiality.

Results: Current study enrollment has surpassed 2000 participating parent-infant triads with an accrual of approximately 100 new families per month. The participation rate of eligible, approached families is approximately 35% with a three year study retention rate of 94%. The longitudinal survey compliance rate is approximately 88%. The direct cost per individual including all sequencing, analysis and other resources required is approximately $4,000. The race and ethnic background of the study cohort is diverse with parents from over 101 countries of origin enabling us to construct novel, population-specific algorithms for the filtering of variants based on sub-population allele frequency. We have constructed a genomic data repository into which we combine data from this and several of our other separate large-scale genomic studies that use similar platforms and which focus on specific conditions such as preterm birth, diabetes mellitus and congenital anomalies (we will generate data on 20-30,000 WGS within several years).

Conclusion: This research creates the infrastructure to integrate clinical and genomic data collected during the infant's first 1,000 days of life. This data provides a large scale model to identify genomic triggers of disease that will establish personalized medicine in pediatrics.

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Implementation of pharmacogenetics into clinical practice

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Advances in pharmacogenetic testing and personalized medicine are beginning to translate into clinical practice. The promise of these advances to improve patient outcomes and reduce cost of care are just being realized, however, an impediment to their rapid adoption has been the novelty of this information and the lack of understanding how to use it in normal clinical practice. As more pharmacogenetic discoveries are made, the information becomes more complex. This is especially true of combinatorial pharmacogenetics which considers multiple genes as they affect response to a particular drug. This lecture will provide a brief overview of pharmacogenetics with special attention to combinatorial pharmacogenetics and as to how this information can be used in clinical decision-making.

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