

“My46,” an innovative web-based tool for management of results return from exome and whole genome sequencing studies

Holly K. Tabor^{1,2}, Joon-Ho Yu², Andrea Civan², Julia Crouch¹, Ellen Kuwana², Margaret J. McMillin², Jacquie Stock³, Karin M. Dent⁴, Nick Anderson^{5,6}, Kimberley Lakes^{7,8}, Bonny Specker⁹, Wylie Burke^{10,11}, Jeff Murray¹², James Swanson^{7,8}, Michael Bamshad^{2,13}

1) Treuman Katz Center for Pediatric Bioethics, Seattle Children’s Hospital, Seattle, WA; 2) Department of Pediatrics, University of Washington, Seattle, WA; 3) Center for Children with Special Needs, Seattle Children’s Hospital, Seattle, WA; 4) Department of Pediatrics, University of Utah, Salt Lake City, UT; 5) Medical Education and Biomedical Informatics, University of Washington, Seattle, WA; 6) Institute of Translational Health Sciences, University of Washington, Seattle, WA; 7) Pediatrics, University of California, Irvine, CA; 8) National Children’s Study Orange County Vanguard Center, Irvine, CA; 9) South Dakota State University, Ethel Austin Martin Program in Nutrition and BYPL Vanguard Center; 10) Department of Bioethics and Humanities, University of Washington; 11) Department of Medicine, University of Washington; 12) University of Iowa, Pediatrics and Polk County IA Study Center ; 13) Genome Sciences, University of Washington, Seattle, WA

Introduction: Whole genome sequencing (WGS) has the potential to identify all variants that influence health-related traits and challenges existing approaches for return of results.

Methods: As a feasibility study for the NCS, we are developing “My46:” a web-based tool that allows research participants to manage return of results from WGS. We developed four modules: 1) “learn the basics” about the genome, including an overview of possible results categories and their potential impact, 2) a tool for selecting preferences for return of results, 3) a platform for reporting results and 4) an instrument for conducting surveys. The preferences tool allows choice of broad return of results categories: disease(s), carrier status for recessive disorders, complex health-related traits, and medication responses. Participants receive available results only in selected categories and can modify preferences at any time. Each module can be customized to fit a study’s specific needs and population.

Results: We are presenting model pages and database structure for My46. Usability testing will proceed in the fall with non-NCS parents. Upon OMB approval, we will test My46 with parents of births in Orange County CA and Brooking County SD Vanguard Centers in the 100 trios WGS project.

Conclusions: “My46” provides participants secure access to their individual genetic results to review at their convenience and as they become available over time. “My46” also facilitates interaction with a genetic counselor via telephone or

live streaming video. These features make “My46” an attractive tool for managing the return of individual research results in WGS studies.