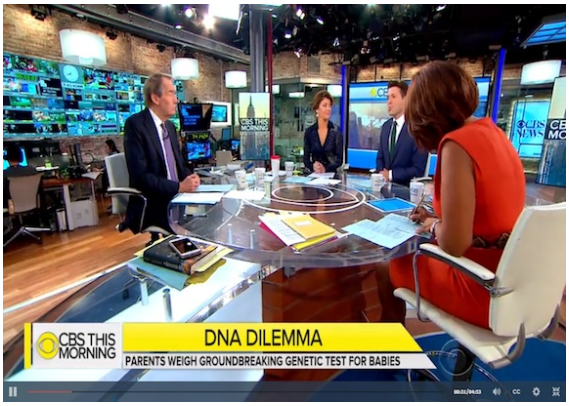
News, Announcements & Other SNP-ets

The Genomes2People Research Program, led by Dr. Robert Green, has had a busy start to fall: kicking off a research collaboration with the U.S. Air Force, speaking at regional and international scientific conferences, interviewing with mainstream media, publishing papers and preparing for our upcoming annual educational genomics event. Read on for more!

Visit our Website

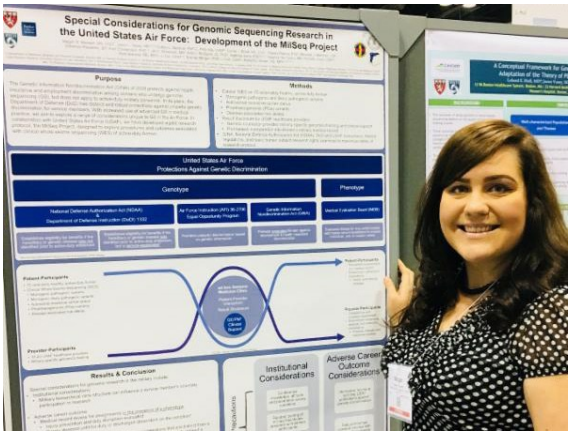
Follow Us On Twitter!

CBS This Morning Highlights the BabySeq Project



On Tuesday, October 24, CBS This Morning highlighted the current progress of our BabySeq Project, a first-of-its-kind randomized clinical trial designed to examine how best to use genomics in clinical pediatric medicine by creating and safely testing methods for integrating sequencing into the care of newborns. Watch the news clip here.

Announcing our Collaboration with the U.S. Air Force



The MilSeq Project, a research collaboration with the U.S. Air Force, is exploring the use of DNA sequencing in routine care of healthy Air Force service members. Now recruiting participants, the partnership was announced during a presentation by Megan Maxwell, MS, CGC, at the recent American Society for Human Genetics meeting where she described the design and early implementation of the study along with special considerations that are unique to genome sequencing in the active-duty military. Read the press release here and GenomeWeb story here.

New MedSeq Publication in Annals

Annals of Internal Medicine ORIGINAL RESEARCH

The Impact of Whole-Genome Sequencing on the Primary Care and Outcomes of Healthy Adult Patients

A Pilot Randomized Trial

Jason L. Vassy, MD, MPH, SM; Kurt D. Christensen, PhD, MPH; Erica F. Schonman, MPH; Carrie L. Blout, MS, CGC; Jill O. Robinson, MA; Joel B. Kirsr, MD; Pamela M. Diamond, PhD; Matthew Lebo, PhD; Kalotina Machini, PhD; Danielle R. Azzariti, MS, CGC; Dmitry Dukhovny, MD, MPH; David W. Bates, MD, MSc; Calum A. MacRae, MD, PhD; Michael F. Murray, MD; Heidi L. Rehm, PhD; Amy L. McGuire, JD, PhD; and Robert C. Green, MD, MPH for the MedSeq Project*

Background: Whole-genome sequencing (WGS) in asymptomatic adults might prevent disease but increase health care use without clinical value.

Objective: To describe the effect on clinical care and outcomes of adding WGS to standardized family history assessment in primary care.

Design: Pilot randomized trial. (ClinicalTrials.gov: NCT 01736566)

Setting: Academic primary care practices.

Participants: 9 primary care physicians (PCPs) and 100 generally healthy patients recruited at ages 40 to 65 years.

Intervention: Patients were randomly assigned to receive a family history report alone (FH group) or in combination with an interpreted WGS report (FH + WGS group), which included monogenic disease risk (MDR) results (associated with Mendelian disorders), carrier variants, pharmacogenomic associations, and polygenic risk estimates for cardiometabolic traits. Each patient met with his or her PCP to discuss the report.

Measurements: Clinical outcomes and health care use through 6 months were obtained from medical records and audio-recorded discussions between PCPs and patients. Patients' health behavior changes were surveyed 6 months after receiving results. A panel of clinician-geneticists rated the appropriateness of how PCPs managed MDR results.

Results: Mean age was 55 years; 58% of patients were female. Eleven FH + WGS patients (22% [95% CI, 12% to 36%]) had new MDR results. Only 2 (4% [CI, 0.01% to 15%]) had evidence of the phenotypes predicted by an MDR result (fundus albipunctatus due to RDH5 and variegate porphyria due to PPOX). Primary care physicians recommended new clinical actions for 16% (CI, 8% to 30%) of FH patients and 34% (CI, 22% to 49%) of FH + WGS patients. Thirty percent (CI, 17% to 45%) and 41% (CI, 27% to 56%) of FH and FH + WGS patients, respectively, reported making a health behavior change after 6 months. Geneticists rated PCP management of 8 MDR results (73% [CI, 39% to 99%]) as appropriate and 2 results (18% [CI, 3% to 52%]) as inappropriate.

Limitation: Limited sample size and ancestral and socioeconomic diversity.

Conclusion: Adding WGS to primary care reveals new molecular findings of uncertain clinical utility. Nongeneticist providers may be able to manage WGS results appropriately, but WGS may prompt additional clinical actions of unclear value.

Primary Funding Source: National Institutes of Health.

Ann Intern Med. 2017;167:xxx-xxx. doi:10.7326/M17-0180 Annals.org

For author affiliations, see end of text.

This article was published at Annals.org on 27 June 2017.

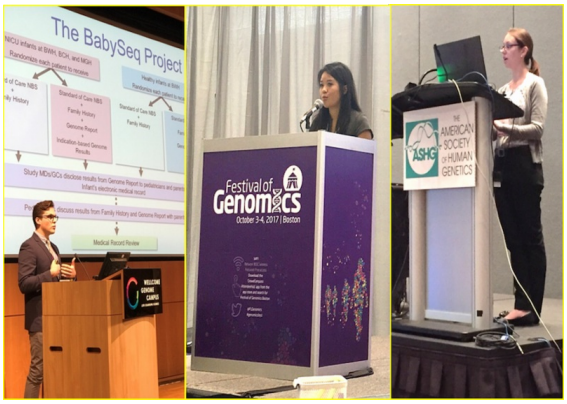
* For members of the MedSeq Project, see the Appendix (available at Annals.org).

Did you know that the MedSeq Project was the first study to comprehensively analyze and report nearly 5,000 disease-associated genes, including monogenic disease risks, carrier variants, pharmacogenomic associations and polygenic risk estimates? And that overall, patients did not exhibit anxiety or distress about their findings? MedSeq is the first-ever randomized clinical trial examining the impact of "end-to-end" whole genome sequencing in healthy adults within the day-to-day practice of medicine. Read the flagship results paper, published in the *Annals of Internal Medicine,* here.

Understand Your Genome® Event November 14th

The 3rd annual Boston Understand Your Genome® Conference is just around the corner! A record number of individuals are signed up for personal genome sequencing and there is an amazing lineup of world class speakers from all facets of the genomics world. A few seats remain for those interested in attending the conference. Sign-up today to experience an incredibly educational day filled with all things genomics! Registration closes soon!

G2P Team Presents at Home and Abroad!



Several G2P team members have recently presented research findings at scientific conferences. To highlight a few, Sheila Sutti, MS, CGC, spoke about the clinical implementation of genetic testing at the Festival of Genomics in Boston. In London, Shawn Fayer, MSc, MS, CGC, spoke about parental interest in genomic sequencing of newborns based on findings from the BabySeq Project at the World Congress on Genetic Counseling. And finally, many talks and posters were shared at the October American Society of Human Genetics meeting. Check out our team's upcoming presentations here!