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News, Announcements & Other SNPets

Jump into spring with the Genomes2People research team! Read on for news about a groundbreaking paper on econogenomics, a new FDA approval for direct-to-consumer genetic testing, and much more.

Visit our Website **Follow us on Twitter!**

Calculating the Cost/Benefit Ratios of Genomic Sequencing

Econogenomics: it's an aspect of the field that is crucially important. What will integrating genomic sequencing into healthcare do to healthcare costs? G2P's Econogenomics Working Group , l ed by Dr. Kurt Christensen, is pioneering research on econogenomics. The first pilot study to look at the cost of using whole genome sequencing in medical care, published in *Genetics in Medicine* , suggests that genomic medicine may not be as costly as some expect it to be. Read the full paper here .

The MedSeq Project was set up specifically to track the outcomes of genomic sequencing in presumably healthy individuals. Read more about MedSeq in our two-part blog series on Medium.

FDA Approves 23andMe to Offer BRCA Testing

Consumers who purchase the 23andMe Health and Ancestry Service will now be told their results for three BRCA mutations most commonly found in the Ashkenazi Jewish population. This is the first instance of a direct-to-consumer genetic testing company being approved by the FDA to report BRCA mutations. Revealing this kind of genetic information to the public without consulting a doctor or genetic counselor is concerning to some experts, but others perceive it as an exciting breakthrough in providing widely accessible health information. Read more about the potential pros and cons and see Dr. Green's comments on the matter here.

The Genomes2People Research Program has conducted one of the only prospective studies of DTC genetic testing customers. See recent publications about DTC testing here .

Meet Megan Maxwell, Genetic Counselor and MilSeq Project Manager

Megan Maxwell is a Genetic Counselor and Project Manager for G2P's MilSeq Project , a collaboration with the U.S. Department of Defense to study whole exome sequencing in the U.S. Air Force. Megan discussed the project and her career as a genetic counselor on the podcast " DNA Today " . Listen to Megan explain her work at G2P and the wide variety of job experiences she has had as a genetic counselor in settings ranging from traditional hospital clinics to sales, education and more. Read more about the MilSeq project here .

The BabySeq Project and Making a Case for Newborn Sequencing

G2P's BabySeq Project, led by Dr. Green and Dr. Alan Beggs of Boston Children's Hospital,

is the first randomized clinical trial to study the effect of using genomic sequencing as a predictive healthcare tool for newborns. Dr. Green visited " The Doctors " talk show to describe the groundbreaking project.

In January, Dr. Green traveled to London to take part in The Great Genome Sequencing Debate, joining Professor Anneke Lucassen, Professor of Clinical Genetics at the University of Southampton to argue in favor of genomic sequencing at birth, against Dr. Kat Arney and Dr. Tara Clancy. Watch the debate here .

Support Genomes2People Research

If you like the work we are doing in accelerating the implementation of genomic medicine, please consider donating to help fund our projects!

Donate

December 2017 Newsletter

News, Announcements & Other SNPets

Season's greetings from Dr. Robert Green and the Genomes2People research team! Read on for an engaging podcast on genomes and sequencing, as well as updates on what we've been up to this fall.

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"Equipped with his five senses, man explores the universe around him and calls the adventure Science."

- Edwin Powell Hubble

Podcast by After On's Rob Reid

Would you want to know your genetic risk for an incurable disease? Would you sequence your newborn infant? Would you share that information with your family, with scientists, or with the general public? Listen to the “After On” podcast by Rob Reid, with Dr. Robert Green, on the pros and cons of having your genome sequenced.

Conference Craze

Seven G2P members presented our science this October at ASHG in Orlando.

In November, Tiffany Nguyen presented an update on REVEAL-SCAN at "Discover Brigham", sharing enrollment progress in our study of amyloid imaging.

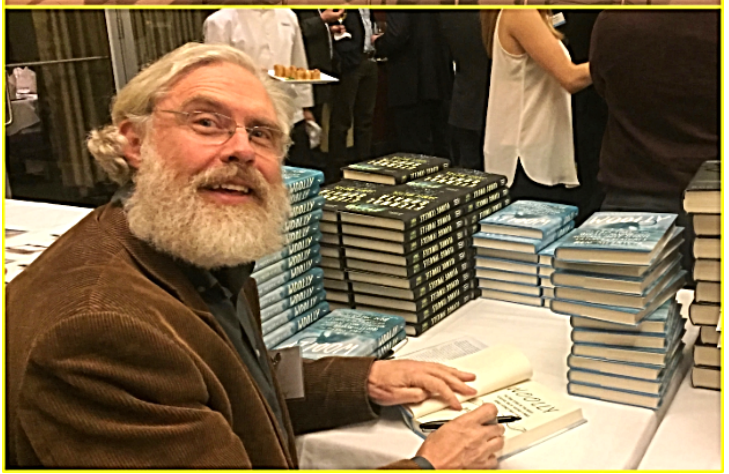
In December, at the Broad Institute, Carrie Blout described our work sequencing active duty military, and Robert Green spoke for the PHG Foundation’s “Healthy Futures: Genomics and Beyond” conference in Cambridge England alongside Professor Dame Sally Davies, Chief Medical Officer for England.

New Publication on Panel Testing vs. Whole Genome Sequencing

We conducted the first head-to-head comparison of whole genome sequencing (WGS) to multi-gene panel testing in patients with cardiomyopathy as part of the MedSeq Project. WGS detected nearly all variants that were identified on panel testing, discovered an additional finding that was missed on the panel, and allowed interrogation for newly discovered disease genes. For more, read the full paper here.

Boston Understand Your Genome ®

Our 3rd annual Understand Your Genome® educational event had a record turnout, including our largest number of genomes sequenced and first diversity genomics panel. The panel, moderated by Jacquelyn Taylor, PhD, RN, discussed the importance of engaging



underrepresented minorities in genomics research with Hannah Valantine, MD, MRCP, Tshaka Cunningham, PhD, and Anthony Johnson, MBA.

At the faculty dinner George Church, Misha Angrist and other speakers signed their books.

We appreciate the support of our conference sponsors: BioNano Genomics, Broad Genomic Services, Dolomite Bio, GenePeeks, Genome, Genome Medical, Genome Web, Helix, Illumina, KolGene, Metabolon, Microbiome Insights, Optalysys, Personalized Medicine Coalition, Qiagen, Qumulo, Sema4, uBiome, and Zymo.

Support Genomes2People Research

Our program is dedicated to accelerating the responsible implementation of genomics into the everyday practice of medicine, but we are entirely dependent upon grant funding which only covers a portion of the costs of our research. Please consider including us in your holiday gifting!

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November 2017 Newsletter

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!

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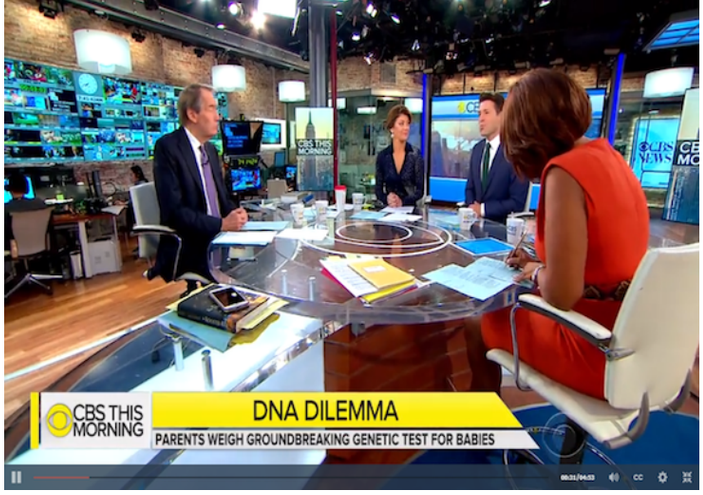
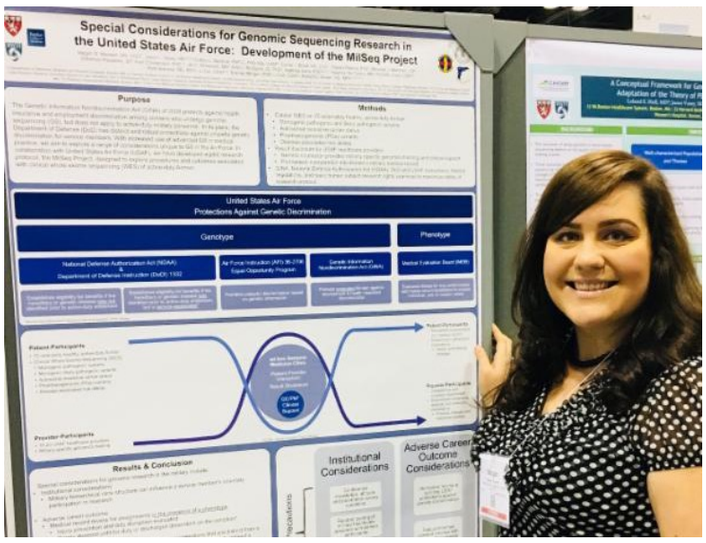
CBS This Morning Highlights the BabySeq Project

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. Krier, 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/M17-0188 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).

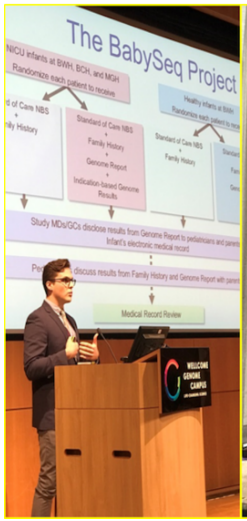
Did you know that the MedSeq Project was the first study to comprehensively analyze and report nearly 5,000 disease-associated genes, including monogenic disease risks, carrier variants, pharmacogenomic associations and polygenic risk estimates? And that overall, patients did not exhibit anxiety or distress 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!

May 2017 Newsletter

G2P
GENOMES *to People*

Greetings!

Thought you might be interested in a recent NYT article describing the impact of genomic information on the life insurance industry, as well as a newly published paper on changes in diet and exercise following direct-to-consumer genetic testing. I also want to invite you to support our staff in their 10K run for G2P research.

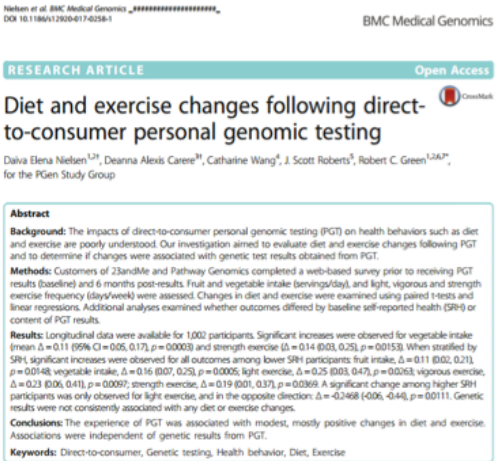
Read on for more!

Thanks,

Robert C. Green, MD, MPH

New Gene Tests Pose a Threat to Insurers

You know you have a genetic risk for Alzheimer's disease. Do you share this information when shopping for life insurance? That's the theme of this recent New York Times article.



Nielsen et al. BMC Medical Genomics
DOI 10.1186/s12920-017-0258-1

BMC Medical Genomics

RESEARCH ARTICLE Open Access

Diet and exercise changes following direct-to-consumer personal genomic testing

Daiva Elena Nielsen[1,2†], Deanna Alexis Carere[3†], Catharine Wang[4], J. Scott Roberts[5], Robert C. Green[1,2,6,7*], for the PGen Study Group

Abstract

Background: The impacts of direct-to-consumer personal genomic testing (PGT) on health behaviors such as diet and exercise are poorly understood. Our investigation aimed to evaluate diet and exercise changes following PGT and to determine if changes were associated with genetic test results obtained from PGT.

Methods: Customers of 23andMe and Pathway Genomics completed a web-based survey prior to receiving PGT results (baseline) and 6 months post-results. Fruit and vegetable intake (servings/day), and light, vigorous and strength exercise frequency (days/week) were assessed. Changes in diet and exercise were examined using paired t-tests and linear regressions. Additional analyses examined whether outcomes differed by baseline self-reported health (SRH) or content of PGT results.

Results: Longitudinal data were available for 1,002 participants. Significant increases were observed for vegetable intake (mean $\Delta = 0.11$ (95% CI = 0.05, 0.17), $p = 0.0003$) and strength exercise ($\Delta = 0.14$ (0.03, 0.25), $p = 0.0153$). When stratified by SRH, significant increases were observed for all outcomes among lower SRH participants: fruit intake, $\Delta = 0.11$ (0.02, 0.21), $p = 0.0148$; vegetable intake, $\Delta = 0.16$ (0.07, 0.25), $p = 0.0005$; light exercise, $\Delta = 0.25$ (0.03, 0.47), $p = 0.0263$; vigorous exercise, $\Delta = 0.23$ (0.06, 0.41), $p = 0.0097$; strength exercise, $\Delta = 0.19$ (0.01, 0.37), $p = 0.0369$. A significant change among higher SRH participants was only observed for light exercise, and in the opposite direction: $\Delta = -0.2468$ (-0.06, -0.44), $p = 0.0111$. Genetic results were not consistently associated with any diet or exercise changes.

Conclusions: The experience of PGT was associated with modest, mostly positive changes in diet and exercise. Associations were independent of genetic results from PGT.

Keywords: Direct-to-consumer, Genetic testing, Health behavior, Diet, Exercise

DTC Genetic Testing and Changes to Diet and Exercise

One of our newest publications, Diet and Exercise Changes Following Direct-to-Consumer Personal Genomic Testing, shows that DTC testing consumers reported improvement in diet and exercise.

Please Support Our G2P 10K Runners!

Members of our research team are filling up their water bottles and lacing up their shoes in preparation for the June Boston Athletic Association 10K Road Race, in which they are raising money to directly support our genomics research efforts. Please make a tax-deductible donation today and help us pursue groundbreaking research pushing the boundaries of genomic medicine!

Donate Here