Personal Genomics: Establishing the Scientific Foundation for Using Personal Genome Profiles for Risk Assessment, Health Promotion and Disease Prevention

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2007: Breakthrough of the Year

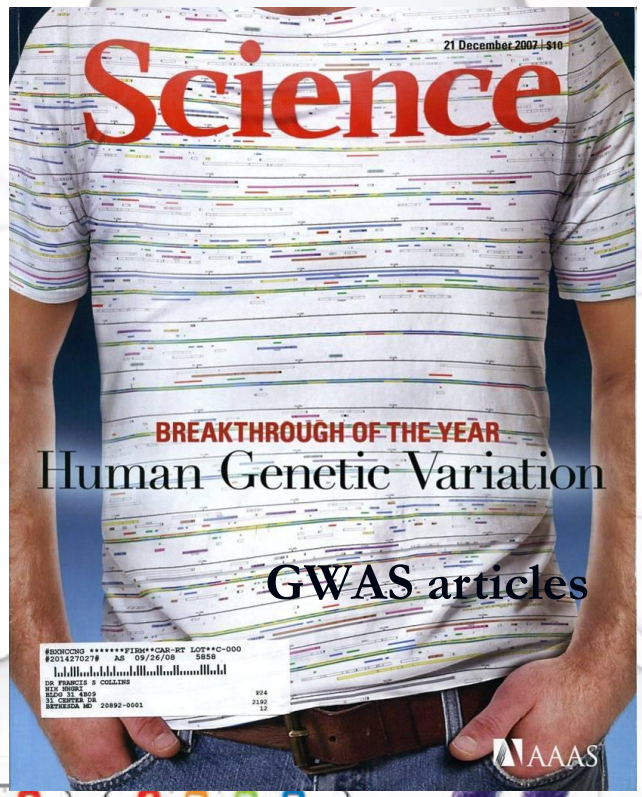
BREAKTHROUGH OF THE YEAR

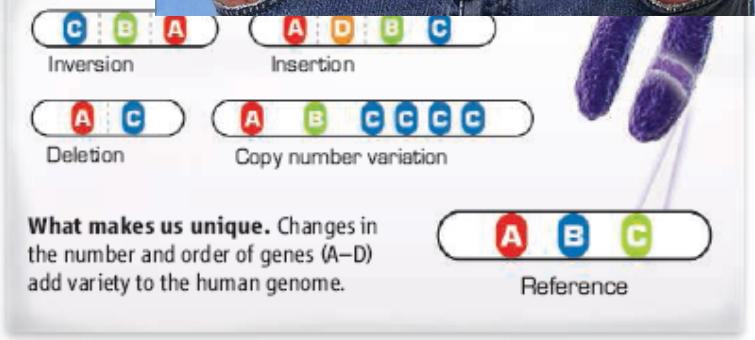
Human Genetic Variation

Equipped with faster, cheaper technologies for sequencing DNA and assessing variation in genomes on scales ranging from one to millions of bases, researchers are finding out how truly different we are from one another

THE UNVEILING OF THE HUMAN GENOME ALMOST 7 YEARS AGO cast the first faint light on our complete genetic makeup. Since then, each new genome sequenced and each new individual studied has illuminated our genomic landscape in ever more detail. In 2007, researchers came to appreciate the extent to which our genomes differ from person to person and the implications of this variation for deciphering the genetics of complex diseases and personal traits.

Less than a year ago, the big news was triangulating variation between us and our primate cousins to get a better handle on genetic changes along the evolutionary tree that led to humans. Now, we have moved from asking what in our DNA makes us human to striving to know what in my DNA makes me me.





Pennisi E, *Science* 2007; 318:1842-43.

2008: Invention of the Year

TIME's Best Inventions of 2008

Invention of the Year

Next

1. The Retail DNA Test

By Anita Hamilton

ARTICLE TOOLS
Print
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AddThis
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Yahoo! Buzz

Before meeting with Anne Wojcicki, co-founder of a consumer gene-testing service called 23andMe, I know just three things about her: she's pregnant, she's married to Google's Sergey Brin, and she went to Yale. But after an hour chatting with her in the small office she shares with co-founder Linda Avey at 23andMe's headquarters in Mountain View, Calif., I know some things no Internet search could reveal: coffee makes her giddy, she has a fondness for sequined shoes and fresh-baked bread, and her unborn son has a 50% chance of inheriting a high risk for Parkinson's disease.

Learning and sharing your genetic secrets are at the heart of 23andMe's service — a $399 saliva test that estimates your predisposition for mor conditions ranging from baldness to blindness. Although 23andMe isn't selling DNA tests to the public, it does the best job of making them acc affordable. The 600,000 genetic markers that 23andMe identifies and

THE BEST INVENTIONS OF THE YEAR

Invention Of the Year

Your genome used to be a closed book. Now a simple, affordable test can shed new light on everything from your intelligence to your biggest health risks. Say hello to your DNA— if you dare

IN DEPTH

Inflated expectorations: At a September "spit party" hosted by 23andMe, invitees supply samples for free genetic testing

And they must be able to analyze genetic data in light of each individual's entire medical history, including lifestyle choices and environmental exposures.

Consider the case of Mike Spear, communications director for Genome Alberta, a Canadian nonprofit. He recently got his genes read by 23andMe. "One of the things that stood out

Time, November 10, 2008

"Scientists Cannot Put the Genie Back in the Bottle"

Perspective

JANUARY 10, 2008

NEJM

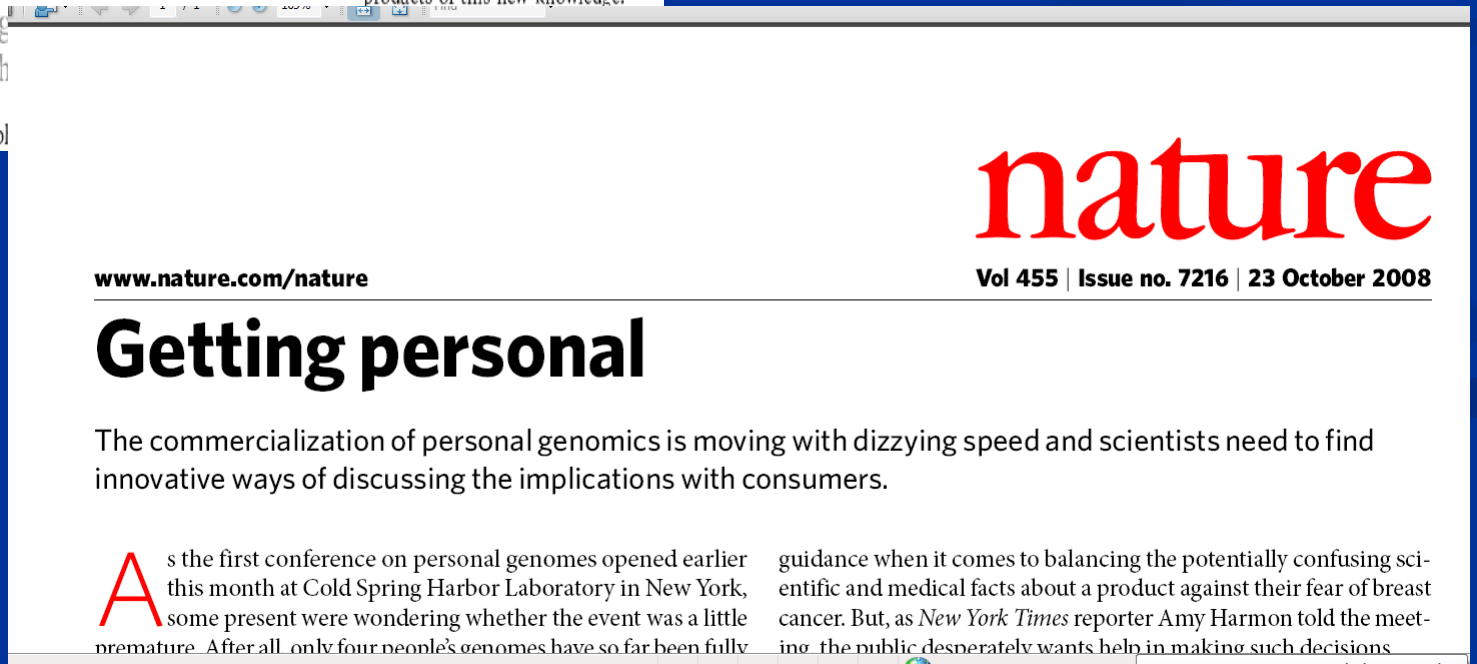
Letting the Genome out of the Bottle — Will We Get Our Wish?

David J. Hunter, M.B., B.S., Sc.D., M.P.H., Muin J. Khoury, M.D., Ph.D., and Jeffrey M. Drazen, M.D.

It may happen soon. A patient, perhaps one you have known for years, who is overweight and does not exercise reg with an analysis of h

single-nucleotide polymorpl

The test undergone by the patient described above is one of the products of this new knowledge.



nature

www.nature.com/nature

Vol 455 | Issue no. 7216 | 23 October 2008

Getting personal

The commercialization of personal genomics is moving with dizzying speed and scientists need to find innovative ways of discussing the implications with consumers.

As the first conference on personal genomes opened earlier this month at Cold Spring Harbor Laboratory in New York, some present were wondering whether the event was a little premature. After all, only four people's genomes have so far been fully

guidance when it comes to balancing the potentially confusing scientific and medical facts about a product against their fear of breast cancer. But, as *New York Times* reporter Amy Harmon told the meeting, the public desperately wants help in making such decisions

The Need for a Translation Roadmap!

COMMENTARY

JAMA Dec 3, 2008

Closing the Evidence Gap in the Use of Emerging Testing Technologies in Clinical Practice

Kathryn A. Phillips, PhD

NEW TESTING TECHNOLOGIES—INCREASINGLY BASED on genomic information—are essential in the shift toward personalized medicine and molecular targeted therapies. Considering the rapid proliferation of new tests, health care insurers and policy makers are interested in assessing evidence about their use and value.

It is critical to build an evidence base to support effective

There is no consensus about optimal testing methods. Guidelines recommend using either immunohistochemistry, with indeterminate results confirmed by fluorescence in situ hybridization (FISH), or FISH to determine HER2 status.[1] Although FISH is a better predictor of response to treatment, immunohistochemistry costs substantially less and is more easily performed in community laboratories.[1]

Despite the clinical success of trastuzumab, there are concerns about the best methods for selecting patients for treatment. The accuracy and interpretation of HER2 tests have

The Evidence Dilemma In Genomic Medicine

We need a roadmap for the appropriate integration of genomic discoveries into clinical practice.

by Muin J. Khoury, Al Berg, Ralph Coates, James Evans, Steven M. Teutsch, and Linda A. Bradley

ABSTRACT: An ongoing dilemma in genomic medicine is balancing the need for scientific innovation with appropriate evidence thresholds for moving technology into practice. The current low threshold allows unsubstantiated technologies to enter into practice, with the potential to overwhelm the health system. Alternatively, establishing an excessively high threshold for evidence could slow the integration of genomics into practice and present disincentives for investing in research and development. Also, variable coverage and reimbursement policies can lead to differential access to technology, exacerbating health disparities. There is an urgent need for a collaborative process for appropriate transition of genomic discoveries from research to practice. [*Health Affairs* 27, no. 6 (2008): 1600–1611; 10.1377/hlthaff.27.6.1600]

PERSPECTIVE

JAMA Dec 10, 2008

The Human Genome And Translational Research: How Much Evidence Is Enough

Given the lack of a robust translational infrastructure, conflic between those developing new technologies and those who r or pay for them seems inevitable.

by Janet Woodcock

ABSTRACT: Multiple new genomic diagnostic tests are currently under d Given the lack of an efficient translational infrastructure, it is not clear how, or bust evidence for their clinical value will be generated. [*Health Affairs* 27, n 1616–1618; 10.1377/hlthaff.27.6.1616]

An Unwelcome Side Effect of Direct-to-Consumer Personal Genome Testing

Raiding the Medical Commons

Amy L. McGuire, JD, PhD
Wylie Burke, MD, PhD

IT IS NOW POSSIBLE FOR INDIVIDUALS TO LEARN ABOUT their genetic susceptibility to dozens of common and complex disorders, such as coronary artery disease,

sel patients accordingly. Physicians are also accustomed to talking with patients about health information disclosed on the Internet or through other media outlets. At the same time, primary care physicians have limited time with patients, face many competing demands,[7] and are poorly reimbursed for time spent counseling patients about preventive care. Patient concerns about direct-to-consumer test results have

Multidisciplinary Translation Sciences of Personal Genomics: Beyond Replication and Biology: Clinical Validity and Clinical Utility?

- Epidemiology
- Clinical trials
- Communication
- Behavioral/social
- Economics
- Outcomes research
- ELSI..

Editorial

Nature **456**, 1 (6 November 2008) | doi:10.1038/456001a; Published online 5 November 2008

My genome. So what?

Research is needed into the way individuals use their genomic information, and into protection from its abuse by others.

Human genome research has proved itself predictably unpredictable. As was widely anticipated, the speed of sequencing has escalated, the pace of linking genes to disease has quickened, and practically anyone can have their genome investigated and fed back to them in electronic format to do with it what they will. In this issue, two groups reveal individual genome sequences of a Yoruba man from Ibadan, Nigeria (see page 53), and of a Han Chinese individual (see page 60) for a cost of less than US$500,000

Nat Genet Aug 2008

Putting science over supposition in the arena of personalized genomics

Colleen M McBride, Sharon Hensley Alford, Robert J Reid, Eric B Larson, Andreas D Baxevanis & Lawrence C Brody

We explore the process of going from genome discovery to evaluation of medical impact and discuss emerging challenges faced by the scientific community. The need to confront these challenges is heightened in a climate where unregulated genetic tests are being marketed directly to the general public[1,2]. Specifically, we characterize

So What Should We Expect from this Meeting?

- Lots of Presentations, Discussions and Hopefully Lively Debate
- Structure
  - I. The Basics
  - II. Credibility of Genetic Associations
  - III, IV. Science of Clinical Validity and Utility
  - V. Case Studies
  - VI. Models for Translation Research
  - VII. Panel Discussion and Next Steps
- Products: A Multidisciplinary Research Agenda and a Scientific Report for Peer Reviewed Publication

Thank you!

- Speakers
- Participants
- Moderators
- Co-sponsors
- NCI
  - Sheri Schully
  - Barbara Guest
  - Camilla Benedicto