

The Public Place in Personal Genomics



M.D.'s, Ph.D.'s, M.D. Ph.D.'s & me

- Confirmed speakers for the event include:
- Bradley Bernstein, M.D., Ph.D.
- Richard Lifton, M.D., Ph.D.
- Lynda Chin, M.D.
- Daniel Kastner, M.D., Ph.D.
- Francis Collins, M.D., Ph.D.
- Amy McGuire, J.D., Ph.D.
- Sean Eddy, Ph.D.
- Maynard Olson, Ph.D.
- Amy Harmon
- David Page, M.D.

- A Decade Later, Genetic Map Yields Few New Cures
- By [NICHOLAS WADE](#)
- Published: June 12, 2010

- Ten years after President Bill Clinton announced that the first draft of the human genome was complete, medicine has yet to see any large part of the promised benefits.
- **The Genome at 10**
- *First of Two Articles*
- For biologists, the genome has yielded one insightful surprise after another. But the primary goal of the \$3 billion Human Genome Project — to ferret out the genetic roots of common diseases like [cancer](#) and [Alzheimer's](#) and then generate treatments — remains largely elusive. Indeed, after 10 years of effort, geneticists are almost back to square one in knowing where to look for the roots of common disease.
- One sign of the genome's limited use for medicine so far was a recent test of genetic predictions for heart disease. A medical team led by Nina P. Paynter of Brigham and Women's Hospital in Boston collected 101 genetic variants that had been statistically linked to heart disease in various genome-scanning studies. But the variants turned out to have no value in forecasting disease among 19,000 women who had been followed for 12 years.

ELSI



RESEARCH

PERSPECTIVE

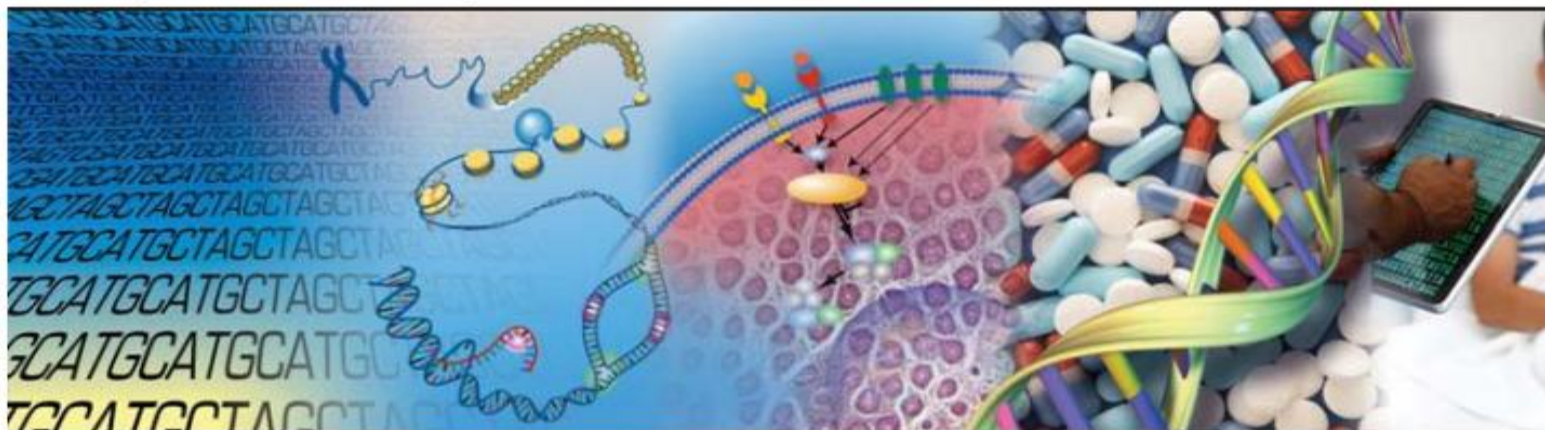
Understanding the structure of genomes

Understanding the biology of genomes

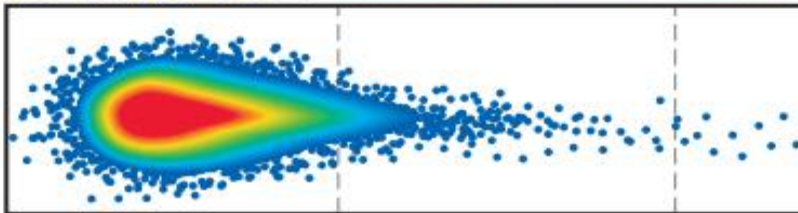
Understanding the biology of disease

Advancing the science of medicine

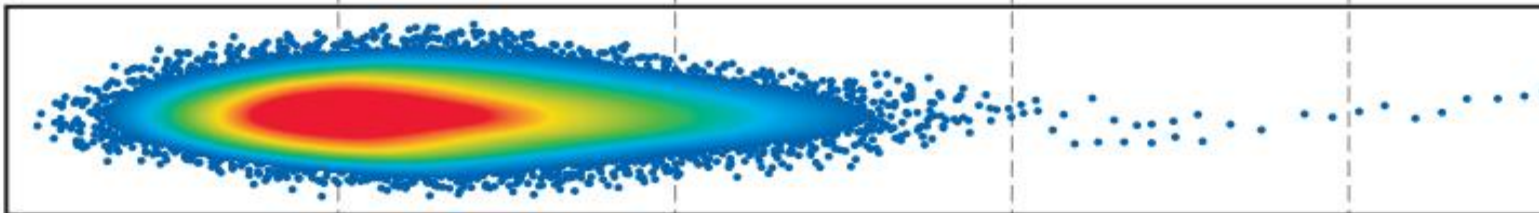
Improving effective health



1990–2003
Human Genome Project



2004–2010



“Community Perspectives”

Do scientists and the public live in different communities?

You talk about the scientific community coming together around the Human Genome Project

Can we merge?

THE DNA AGE

After DNA Diagnosis: 'Hello, 16p11.2. Are You Just Like Me?'



Samantha Napier, 14, left, and Taygen Lane, 4, share a rare genetic mutation.

Why bother??

THE DNA AGE

After DNA Diagnosis: 'Hello, 16p11.2. Are You Just Like Me?'



Samantha Napier, 14, left, and Taygen Lane, 4, share a rare genetic mutation.

Katharine Moser – tested positive for Huntington’s gene mutation



People take genomics personally

- Americans' Concern about the Privacy of Their Genetic Information Reaches New High
- Cambridge, Mass., January 10, 2011 - A report just released by Cogent Research, in partnership with CAHG, reveals that Americans are more concerned than ever about the privacy of their genetic information. Furthermore, this concern is increasingly inhibiting the likelihood that they will consider having a molecular diagnostic test.
- These and other findings are explored in more detail in the 5th edition of the Cogent Genomics, Attitudes & Trends study (CGAT™ 2010).
- The CGAT™ study, based on a nationally representative survey of 1,000 Americans, reveals that the proportion of Americans who are concerned about how their genetic information would be stored and who would have access to that information, has climbed from 65% in 2006 to an all-time high of 71% in 2010.

Carletta Tilousi, Havasupai Tribe



“Help!!! The Gov’t Has My DNA”



Who, what why when & how?

“Conducting public outreach.

Education programmes are needed to promote lifelong public understanding and awareness of the role of genomics in human health and other areas.”

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EDITORIAL:

Making Data Maximally Available
 Hanson, et al.
 Science 11 February 2011: 649.
 DOI:10.1126/science.1203354

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Kerri Adams: BRAF-positive melanoma



Randy Williams



Mark Bunting



B-RAF resistance paper in Nature

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LETTER

doi:10.1038/nature09626

Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation

Ramin Nazarian^{1,2*}, Hubing Shi^{1,2*}, Qi Wang^{1,2}, Xiangju Kong^{1,2}, Richard C. Koya^{2,3}, Hane Lee^{2,4}, Zugen Chen^{2,4}, Mi-Kyung Lee^{1,2}, Narsis Attar^{2,5}, Hooman Sazegar^{2,5}, Thinle Chodon^{2,5}, Stanley F. Nelson^{2,4,6}, Grant McArthur⁷, Jeffrey A. Sosman⁸, Antoni Ribas^{2,3,5} & Roger S. Lo^{1,2}

Activating B-RAF(V600E) (also known as BRAF) kinase mutations occur in ~7% of human malignancies and ~60% of melanomas¹. Early clinical experience with a novel class I RAF-selective inhibitor, PLX4032, demonstrated an unprecedented 80% anti-tumour response rate among patients with B-RAF(V600E)-positive melanomas, but acquired drug resistance frequently develops after initial responses². Hypotheses for mechanisms of acquired resistance to B-RAF inhibition include secondary mutations in B-RAF(V600E),

There were no secondary mutations in the drug target *B-RAF* (V600E) observed on bi-directional Sanger sequencing of all 18 *B-RAF* exons in 15 M229 R (R1–R15), two M238 R (R1 and R2), and one M249 R (R4) acquired resistant sub-lines (Supplementary Table 1 and Supplementary Fig. 3a, left column). Based on Sanger sequencing, this lack of secondary *B-RAF* (V600E) mutation along with retention of the original *B-RAF* (V600E) mutation was confirmed in 16/16 melanoma tumour biopsies (from 12 patients) with clinically acquired resistance

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