

# Personalized Health Care

“Your life depends on the secrets of your DNA:  
Are you ready?”

Abdallah S Daar

McLaughlin-Rotman Centre for Global Health,  
University Health Network and University of  
Toronto

# Desmond Tutu's genome sequenced as part of genetic diversity study

Archbishop Desmond Tutu has had his genome sequenced in research to reveal the true breadth of human genetic diversity

**Ian Sample**, science correspondent

[guardian.co.uk](http://guardian.co.uk), Wednesday 17 February 2010 18.02 GMT

[Article history](#)



The genomes of Desmond Tutu and four bushmen have been added to a database

# Major Developments on Road to Modern Personalized Health Care

McLaughlin-Rotman Centre  
FOR GLOBAL HEALTH

- Human Genome Project: ? the **most successful** research enterprise ever
- Advances in technology (1000\$ sequencing; 200\$)
- Int'l HapMap Project: defined neighborhoods of packs of SNPs that travel together
- Genome Wide Association Scans “few, if any, similar outbursts of discovery in the history of medical research (*NEJM*); human genetic variation studies: “Breakthrough of the Year” for 2007 (*Science*)
- Proteomics; other –omics; systems biology
- Understanding alternate splicing (one brain gene codes for 30,000 proteins)
- Personal Genome Project; Cancer Genome Project; Epigenetics; Microbiome
- Copy Number Variations
- Public Health Agencies focus on public health genomics
- FDA and industry interest
- Stem cells (iPS; personalized stem cell therapy)
- **Francis Collins**

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## Books and Arts

*Nature* **463**, 298-299 (21 January 2010) | doi:10.1038/463298a; Published online 20 January 2010

### Vision of a personal genomics future

Abdallah S. Daar<sup>1</sup>

**The director of the US National Institutes of Health, Francis Collins, calls for a revolution in personalized medicine. Such advances should be shared beyond the developed world, says Abdallah S. Daar.** [▲ Top](#)






#### BOOK REVIEWED

***The Language of Life: DNA and the Revolution in Personalized Medicine***

by Francis S. Collins

*Harper/Profile*: 2010. 368 pp/288 pp. \$26.99/£15

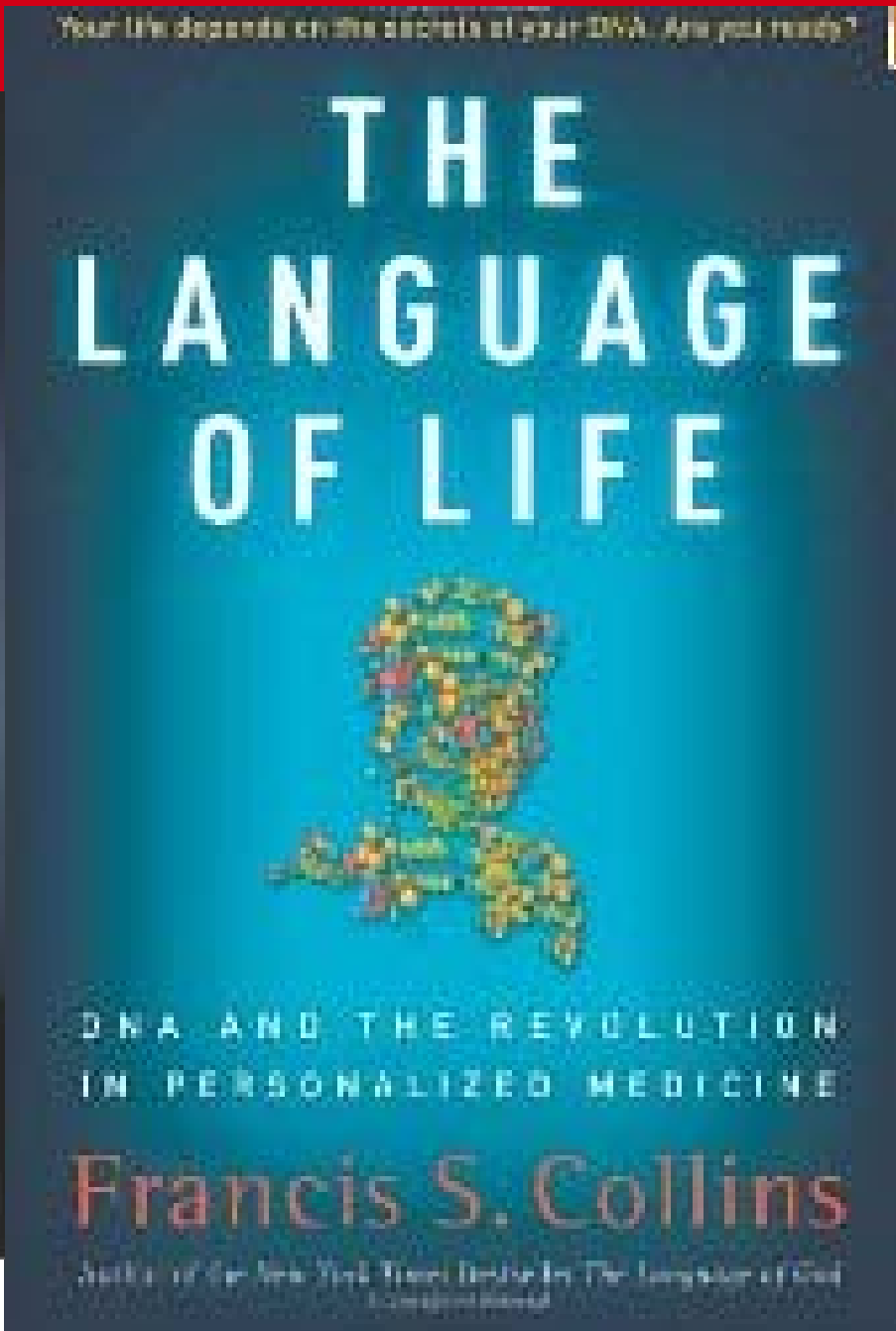
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# “The Language of Life: DNA and the Revolution in Personalized Medicine”.

Francis Collins

- Ch.1: The Future Has Already Happened
- 2: When Genes Go Wrong, It Gets Personal
- 5: What’s Race Got to Do With It?
- 7: Genes and the Brain
- 8: Genes and Aging
- 9: The Right Drug at the Right Dose for the Right Person
- Each chapter ends with “What You Can Do Now To Join The Personalized Medicine Revolution”

- Genetic medicine is at a tipping point. Era of personalized medicine has arrived.
- Genes are generally not destiny, especially for common conditions like heart disease, diabetes or cancer
- Genes load the gun, and environment pulls the trigger
- There are virtually no conditions for which heredity does not play a role
- 60-70 % of one's adult body weight is determined by genes (so modify diets early)
- Family history is a free genetic test
- Whether now or in the near future, it is time to test your own DNA-there are dramatic anecdotes suggesting potential info of considerable impact

# Defining What is Involved in Personalized Medicine / Care

- Application of science of genomics (variation), proteomics, molecular pathways, bioinformatics, etc, for (individual)
- Susceptibility
- Prevention
- Diagnosis (including point-of-care)
- Care/ therapy (Phgx);
- Lifestyles: diet; physical activity
- Prognosis
- Improve health outcomes
- Reduce health care costs



# We Have Always Had Personalized Medicine, So What's New?

1. Willingness to share info and partially forego confidentiality (Venter; Watson; Collins, Desmond Tutu, Pinker . Personal Genome Project (Church)
2. Understanding disease at molecular level, with some dramatic interventions
3. Public is more informed and interested: Desire to Know
4. Better understanding of risk factors for epidemic of chronic diseases; Prevention is becoming popular; prognosis linked to better health behaviour (attention to weight; nutrition; cholesterol; exercise; smoking; alcohol; etc): from private care to public health care

# We Have Always Had Personalized Medicine, So What's New?

5. More disposable income means better focus on health and “recreational genomics”
6. With more expensive drugs, more attention to “rescuing” drugs
7. Insurance may pay for DNA tests
8. Internet, direct-to-consumer (DTC) genetic testing companies (23andMe was 2008 Time Magazine’s Invention of the Year- 399\$; Navigenics, DeCODEme, etc) are here, but need oversight
9. Industry, regulatory agencies (FDA) interest

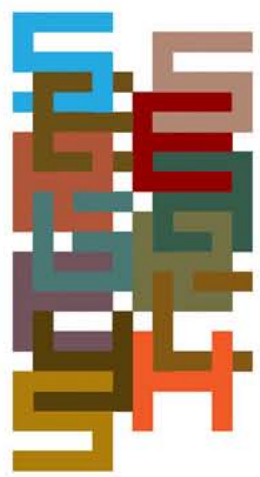
News Bike's Design

# MAGAZINE

PLAY 18.09

## Sergey Brin's Search for a Parkinson's Cure

By Thomas Goetz June 22, 2010 | 12:00 pm | Wired July 2010



Buried deep within each cell in Sergey Brin's body—in a gene called LRRK2, which sits on the 12th chromosome—is a genetic mutation that has been associated with higher rates of Parkinson's.  
Illustration: Rafa Jenn

Several evenings a week, after a day's work at Google headquarters in Mountain View, California,

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- LRRK2 mutation increases his risk of Parkinson's from 1% to 50%
- Wants to reduce that to 10% through lifestyle modifications; and (funding) research
- A more Googley kind of science: “collect data first, then hypothesize, and then find the patterns that lead to answers. And he has the money and the algorithms to do it”
- PERSONALIZED INVESTIGATION

- Genomics will impact all areas of public health (infectious diseases, environmental and occupational health, chronic diseases, maternal and child health, etc).
- Address growing schism between public health and medicine by focusing on prevention and early intervention
- **Translation Highway**
- CDC's EGAPP initiative (Evaluation of Genomic Applications in Practice and Prevention): an independent multidisciplinary working group making evidence-based recommendations on pharmacogenomics and other genomic applications in practice
- US Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS)

# GRaPH-Int

Genome-based Research and Population Health International Network

About US

The Enterprise

Areas of Activity

Organizations

Network News

Home >>The Enterprise >> Diagram

The enterprise translates genome-based science and technology into improvements in population health. The functions and activities shown in blue define the scope of the enterprise defined at the **Bellagio meeting**. Yellow represents the generation of knowledge through research, and green represents all the activities, people, institutions and views that make up society. The ultimate goal is to improve population health.

*\*\*\*This web page has been formatted in a manner that allows a person to use the enterprise diagram as a navigation tool. A number of components (hot spots) within the diagram below will take a person to a sub-page specific to that topic.\*\*\**

## Member's Network

### Videos

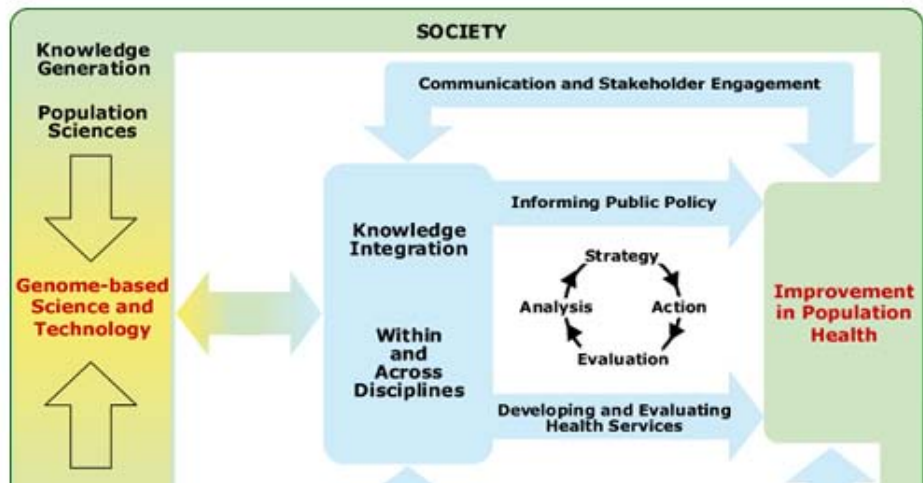


Welcome remarks from the Chairman of Graph-Int

### Service



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- Predict
  - response
  - dosage
- Eliminate
  - adverse reactions
  - unnecessary treatments
- Enhance efficacy
- Resuscitate old drugs
- Rationalize clinical trials
- Some examples: Warfarin, Azathioprine and Mercaptopurine, Iressa, Abacavir, Clopidogrel, etc

# Examples of Potential Therapeutic Breakthroughs

- VX-770 (Cystic fibrosis potentiator): results of initial trial “astounding”
- Macular degeneration: two different genetic mutations (inflammatory pathway) account for 80% of risk, combined with smoking and obesity; now anti-inflammatories, omega-3 fatty acids, and gene therapy
- Selzendry (Mariviroc) binds to CCR5, approved by FDA 2007: considerable promise for treatment of HIV infection



# Oncology: Cancer is a Disease of the Genome

- Gleevec
- BRCA
- Iressa
- PLX 4023

TARGET CANCER

## New Drugs Stir Debate on Rules of Clinical Trials



Monica Almeida/The New York Times, left

**Two Cousins, Two Paths** Thomas McLaughlin, left, was given a promising experimental drug to treat his lethal skin cancer in a medical trial; Brandon Ryan had to go without it.

By AMY HARMON

# Oncology: Do We Still Need Randomized Control Trials?

- PLX 4023: vastly better responses (in 50% with B-RAF mutation), vastly less toxic in advanced melanoma compared to standard chemo agent dacarbazine
- Fundamentally different type of drug, NEJM “a major breakthrough”; Biology understood
- “Excuse me, but if it was your own life on the line, Doctor, would you take dacarbazine?”
- Patient: “It doesn’t make sense to say we want you for a statistic instead of giving them a chance at life”

- Susceptibility genes (TB, Flu)
- Vaccinogenomics (see Joly , McLellan and Knoppers 2010)
- Personal microbiome
- Drug treatment e.g. CCR5 blocking drugs
- Pharmacogenomics (e.g. Abacavir)
- Point of care diagnostics
- Genomes of parasites & their vectors (targets)

# Chronic Non-communicable Disease

- For each disease, specific genetic (most are surprises) and environmental risk factors exist, and are rapidly being identified; each common variant contributes only small amount of risk (missing heritability)
- Discoveries provide powerful new insights into both treatment and prevention
- Knowledge allows adjustment of lifestyle (dramatic results from Diabetes Prevention Program) and medical surveillance to prevent illnesses or to catch them early
- Classification of disease will need serious revisio
- Genes coding for nicotine receptors also predispose to addiction and to ca lung

- Be informed
- Plan for the long term, starting with public and particularly in medical school
- Avoid both “lost in translation” and “premature translation”; systematic approach , construct a “translation highway”:
- Evaluate the emerging candidate applications; build the evidence base for their appropriate utilization in practice
- Collaborative framework that rewards scientific innovation and appropriate clinical applications.
- Policy framework for accurate laboratory testing, truth in advertising, and protection from untoward psychological or social effects

- Don't count on primary care physician to be well enough informed to give you advice
- Don't get tested until you understand all the possible consequences
- Since none of us can choose our DNA, it should not be used to discriminate against us- any more than ....the color of our skin
- Sequencing the human genome was substantially easier than than getting a well-crafted bill (GINA) passed by both houses of Congress

- Advice from Wayne Gretsky's Father: "Skate where the puck is going to be"
- **PERSONALIZED MEDICINE REQUIRES PERSONALIZED RESPONSIBILITY**