

Health and Illness in the Age of Genomics

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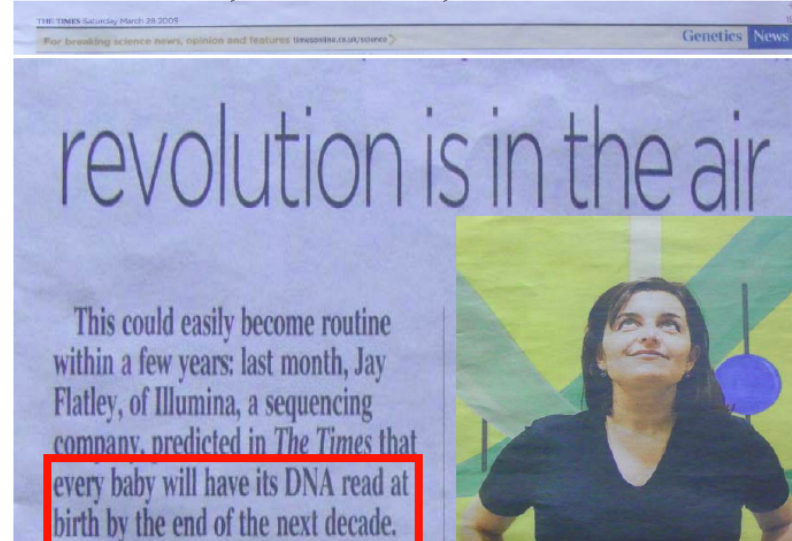
November 2010



VISIONS OF THE FUTURE

In this new three-part series, leading theoretical physicist and futurist Dr Michio Kaku explores the cutting edge science of today, tomorrow, and beyond. He argues that humankind is at a turning point in history. In this century, we are going to make the historic transition from the 'Age of Discovery' to the 'Age of Mastery', a period in which we will move from being passive observers of nature to its active choreographers. This will give us not only unparalleled possibilities but also great responsibilities.

The Times, March 28, 2009



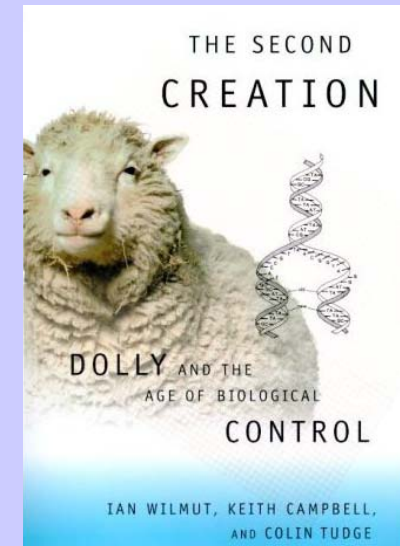
2. THE BIOTECH REVOLUTION

Genetics and biotechnology promise a future of unprecedented health and longevity: DNA screening could prevent many diseases, gene therapy could cure them and, thanks to lab-grown organs, the human body could be repaired as easily as a car, with spare parts readily available. Ultimately, the ageing process itself could be slowed down or even halted.

But what impact will this have on who we are and how we will live? And, with our mastery of the genome, will the human race end up in a world divided by genetic apartheid?

The Age of Biological Control - Ian Wilmut (2000)

- “Until the birth of Dolly, scientists were apt to declare that this or that procedure would be ‘biologically impossible’-- but now that expression ...seems to have lost all meaning. In the 21st century and beyond, human ambition will be bound only by the laws of physics, the rules of logic, and our descendents’ own sense of right and wrong. Truly, Dolly has taken us into *the era of biological control*”
- “This means that we can no longer assume that the biological itself’ will impose limits on human ambitions. As a result, humans must accept much greater responsibility toward *the realm of the biological, which has, in a sense, become a wholly contingent condition*”

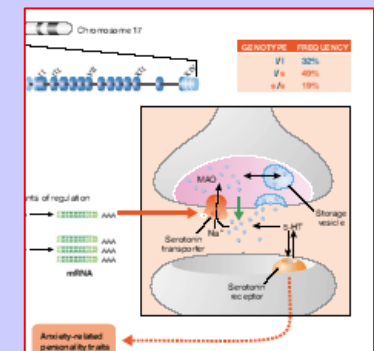
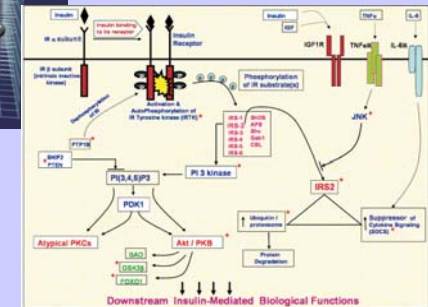
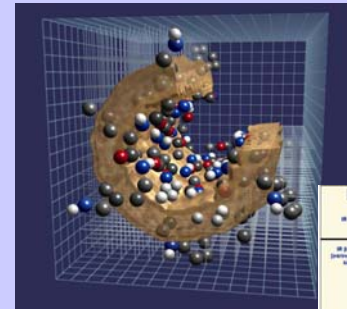


CONTINGENT

- A. *adj.* ...
- 2.
 - a. Liable to happen or not; of uncertain occurrence or incidence.
 - b. Incidental (*to*).
- 4. Happening or coming by chance; not fixed by necessity or fate; accidental, fortuitous.
- 5. Not determined by necessity in regard to action or existence; free. *Obs.*
- 6. Subject to or at the mercy of accidents; liable to chance and change.
- 7. *Metaph.*
 - a. Not of the nature of necessary truth; true only under existing conditions.
 - b. That does not exist of itself, but in dependence on something else.
 - c. Non-essential.
- 8. Dependent for its occurrence or character *on or upon* some prior occurrence or condition.

Biological control?

- Scientific hubris of course
 - much remains 'biologically impossible'
 - 'nature' says no as often as yes
- But points to a mutation in our very idea of life
 - Life as mechanism, bodies and brains as engineerable apparatus
 - Molecular gaze of biology
- Beyond the binary of the normal and the pathological
 - no-one is normal, normality and pathology explained in the same terms, no such thing as normal body, brain, genome, scan, aging...
- An age of **control** over our biology?
 - Bioprediction – to know and control our biofutures.



China overtakes India in drug testing

By Andrew Jack in London and Amy Yee in New Delhi, Published: August 27 2007 23:31

China has overtaken India as one of the fastest-growing locations for drug trials, in a fresh sign of the importance of the world's most populous country to the pharmaceutical industry.

An analysis by the Financial Times of data on www.clinicaltrials.gov, one of the most comprehensive websites where researchers register their studies, shows that China has 274 clinical trials under way, compared with 260 in India.

That site also indicates that China now has a cumulative total of 510 completed or ongoing trials compared with 471 in India, which had until recently been ahead on both measures. The trend reflects intensifying interest by the healthcare sector in China, which is growing rapidly as a result of rising income and expanding health coverage and is already forecast to be the world's fifth-largest pharmaceuticals market by 2010.

Daniel Vasella, chief executive of Novartis, the Swiss pharmaceutical company, warned earlier this month that he was likely to switch substantial future funding that could have gone to India to other countries, including China, because of a recent court ruling on patents.

As they seek to reduce the escalating costs and speed up the conduct of the clinical trials necessary to win regulatory approval for new medicines, drug companies are increasingly shifting tests from the US and western Europe to eastern Europe, Latin America and Asia.

India and China have both received increased attention by pharmaceutical companies in recent years, reflecting a strong medical infrastructure, substantially lower costs and the relative ease of recruiting patients with diseases under investigation – which allows trials to be launched more rapidly.

Last updated at: (Beijing Time) Thursday, March 11, 2004

China launches first biobank

China recently launched its first biobank, a database with information on people's medical history, lifestyle, occupation and blood sample for DNA analysis in Guangzhou, capital of South China's Guangdong Province.

The Kadoorie Biobank Study in China 中英合作慢性病前瞻性研究项目

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The Kadoorie Biobank Study in China (KBSC) is a prospective health study investigating chronic diseases, such as stroke, heart disease, cancer, diabetes and chronic respiratory diseases. 513,000 people aged 30-79 were recruited from 10 areas across China. During the study, participants consented to take part, had a blood sample taken, underwent physical examination, completed a questionnaire. Participants will be closely monitored over the coming decades for hospital admissions and deaths. More details about the study are available in [About the study](#). The data and resources available, including biological samples, can be found in [Resources](#).

The KBSC is a collaborative project between the [Clinical Trial Service Unit & Epidemiology Unit \(CTSUEU\)](#) at the [University of Oxford](#) and the [Chinese National Center for Disease Control and Prevention \(China CDC\)](#). The study was set up with financial support from the [Kadoorie Foundation](#), Hong Kong, and additional funding has come from the [Wellcome Trust](#), the [Medical Research Council \(UK\)](#) and the [British Heart Foundation](#). The study was previously called the Kadoorie Study of (KSCDC).

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中国人群遗传多样性数据库
Genetic Diversity Database of Chinese Population

A brief introduction

China is a country with long history and broad areas. The population of it is nearly 1,300,000,000 and is divided into 56 ethnic groups. Those minorities have various origins, settled areas, different languages, cultures and customs. Among of them, there are even several groups living isolated from others. Recent researches have found that genetic diversities between different groups are strong related to human health and diseases. Research on genetic diversity has become an important fields for a comprehensive understanding of human genome and its function. The research result is very significant for basic research in medical genetics, related fields with human health, individual identification in medical jurisprudence, human origin, evolution and migration.



Our country is well known as a nation with abundant human genetic resources. Genetic diversity between different groups has long been an important part in Chinese human genome research. Under the support of the government and the efforts that the scientists make, genetic diversity research in China has achieved great success. A lot of information and

“China Uncorks the Gene in a Bottle”

Far Eastern Economic Review 03/22/2001 by David Murphy

*“So what has China achieved in the life sciences so far?
A good place to begin answering that question is the
Beijing Genomics Institute...”*



The 1000 Genomes Project

*a catalogue of
human
polymorphism
created using
next generation
sequencing*



nature

Vol 456 | 6 November 2008 | doi:10.1038/nature07484

ARTICLES

The diploid genome sequence of an Asian individual

Jun Wang^{1,2,3,4*}, Wei Wang^{1,3*}, Ruiqiang Li^{1,3,4*}, Yingrui Li^{1,5,6*}, Geng Tian^{1,7}, Laurie Goodman¹, Wei Fan¹, Junqing Zhang¹, Jun Li¹, Juanbin Zhang¹, Yiran Guo^{1,7}, Binxiao Feng¹, Heng Li^{1,8}, Yao Lu¹, Xiaodong Fang¹, Huqing Liang¹, Zhenglin Du¹, Dong Li¹, Yiqing Zhao^{1,7}, Yujie Hu^{1,7}, Zhenzhen Yang¹, Hancheng Zheng¹, Ines Hellmann⁹, Michael Inouye⁹, John Pool⁹, Xin Yi^{1,7}, Jing Zhao¹, Jinjie Duan¹, Yan Zhou¹, Junjie Qin^{1,7}, Lijia Ma^{1,7}, Guoqing Li¹, Zhenao Yang¹, Guojie Zhang^{1,7}, Bin Yang¹, Chang Yu¹, Fang Liang^{1,7}, Wenjie Li¹, Shaochuan Li¹, Dawei Li¹, Peixiang Ni¹, Jue Ruan^{1,7}, Qibin Li^{1,7}, Hongmei Zhu¹, Dongyuan Liu¹, Zhike Lu¹, Ning Li^{1,7}, Guangwu Guo^{1,7}, Jianguo Zhang¹, Jia Ye¹, Lin Fang¹, Qin Hao^{1,7}, Quan Chen^{1,5}, Yu Liang^{1,7}, Yeyang Su^{1,7}, A. san^{1,7}, Cuo Ping^{1,7}, Shuang Yang¹, Fang Chen^{1,7}, Li Li¹, Ke Zhou¹, Hongkun Zheng^{1,4}, Yuanyuan Ren¹, Ling Yang¹, Yang Gao^{1,6}, Guohua Yang^{1,2}, Zhuo Li¹, Xiaoli Feng¹, Karsten Kristiansen¹, Gane Ka-Shu Wong^{1,10}, Rasmus Nielsen⁹, Richard Durbin⁹, Lars Bolund^{1,11}, Xiuqing Zhang^{1,5}, Songgang Li^{1,5}, Huanming Yang^{1,2,3} & Jian Wang^{1,2,3}

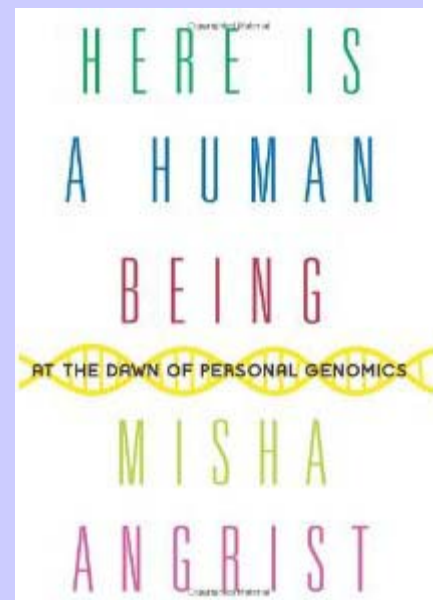
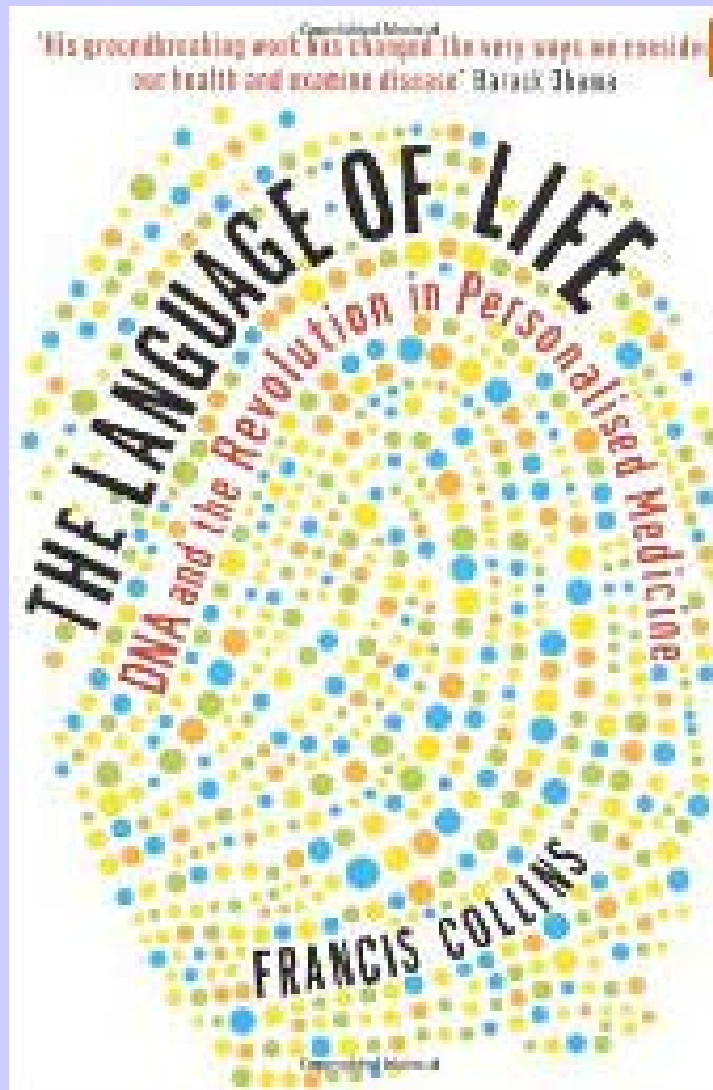
Here we present the first diploid genome sequence of an Asian individual. The genome was sequenced to 36-fold average coverage using massively parallel sequencing technology. We aligned the short reads onto the NCBI human reference genome to 99.97% coverage, and guided by the reference genome, we used uniquely mapped reads to assemble a high-quality consensus sequence for 92% of the Asian individual's genome. We identified approximately 3 million single-nucleotide polymorphisms (SNPs) inside this region, of which 13.6% were not in the dbSNP database. Genotyping analysis showed that SNP identification had high accuracy and consistency, indicating the high sequence quality of this assembly. We also carried out heterozygote phasing and haplotype prediction against HapMap CHB and JPT haplotypes (Chinese and Japanese, respectively), sequence comparison with the two available individual genomes (J. D. Watson and J. C. Venter), and structural variation identification. These variations were considered for their potential biological impact. Our sequence data and analyses demonstrate the potential usefulness of next-generation sequencing technologies for personal genomics.

Genomic medicine

PPP: personalised, predictive, preventive

- Leroy Hood, 1992:

“The genome project in the twenty-first century will have a profound impact on medicine, both for diagnosis and therapy ... Perhaps the most important area of DNA diagnostics will be the identification of genes that predispose individuals to disease. However, many such diseases – cardiovascular, neurological, autoimmune – are polygenic; they are the result of the action of two or more genes. Human genetic mapping will permit the identification of specific predisposing genes and DNA diagnostics will facilitate their analysis in many different individuals ... Perhaps in twenty years [he was writing in 1992] it will be possible to take DNA from newborns and analyze fifty or more genes for the allelic forms that can predispose the infant to many common diseases... For each defective gene there will be therapeutic regimens that will circumvent the limitations of the defective gene. Thus medicine will move from a reactive mode ... to a preventive mode. Preventive medicine should enable most individuals to live a normal, healthy, and intellectually alert life without disease.



Has the
genomics
revolution
finally arrived?



Direct-to-consumer personal genetic profiling

Company	Example product	Price	Details
23andMe	Health Edition	\$429	"Find out if you carry inheritable markers for diseases such as breast cancer, cystic fibrosis, and Tay-Sachs...Learn your genetic risk for type 2 diabetes, Parkinson's disease, and other conditions." ⁴²⁰
deCODEme	Complete Scan	\$2000	"Calculate your genetic risk for 51 conditions..." ⁴²¹
Genetic Health	Premium Male	£825	"These are our most comprehensive test and includes all the other tests in our range... Evaluates the risk of prostate cancer as well as the risk for thrombosis, osteoporosis, metabolic imbalances of detoxification and chronic inflammation. It also evaluates the risk profile of the most common cardiovascular diseases..." ⁴²²
Graceful Earth	Alzheimer's genome test	\$280	"Check your future susceptibility BEFORE symptoms occur... Pre-emptive insight into one's genetic predisposition can empower and allow for pro-active prevention." ⁴²³
Navigenics	Health Compass	Varies	"Knowing your genetic predispositions for important health conditions and medication reactions can help motivate you to take steps towards a healthier life. By gaining insight into these risks, you can plan for what's important." ⁴²⁴

Knowing my very own future?

- Scan me
- Decode me
- The book of me
- Personalise me
- Kno(w)me
- Commodification of 'personal risk'
- Individualised biological prudence

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THE BOOK OF ME
 If you could see into your future, would you want to? If you could know whether you're going to contract Alzheimer's, or if you're likely to battle cancer or die of heart disease, would you want to? Last summer Richard Powers decided he did and became one of nine people on earth to have his entire genome sequenced. Here, a glimpse into his—and your—future
 By Richard Powers; Photograph by Kevin Van Aelst

I come from a long line of folks, on my mother's side, with congenital difficulty making choices. My father's family, on the other hand, are born snap deciders. This time the paternal genes won out, and half an hour after reading the invitation, I was on board.

So I went shopping. A day online gave me my first taste of the bewildering range of consumer genetic products. There was Family Tree DNA, specializing in tracing genetic genealogies. There was DNA Direct, whose Web site asked, "Do you have a chronic, undiagnosed condition? It could be genetic." For \$260, I could get tested for cystic fibrosis; for \$370, I could learn whether I'm at elevated risk of developing type 2 diabetes. Then there was Iceland-based deCODEme ("This is myCODE"), which could calculate my risks for twenty-five genetic maladies in one \$985 package.

But why stop with just a few disease tests? As I always say, in for a few plot complications, in for the whole story.

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"We have nothing but praise for the kind, courteous and sympathetic way in which we were treated. We feel it was well worth the money!"
 John and Eunice Malloch

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 Cancer
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 Optimised Health and Performance Screen
 Anti-Ageing Profile
 Digestive efficiency
 How is stress affecting you?

Top-ten targeted health screens
Cancer risk / early detection
 providing very early indications of any potential cancerous activity and an assessment of your risk factors.
Heart disease risk / early detection
 providing very early indications of any potential heart problems and an assessment of your risk factors
Optimised health and performance

How is stress affecting you?
 to help understand the impact of stress on your body and identify ways to manage it better.
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 blood and urine tests investigating for the presence of the main pollutants and potentially toxic chemicals which are widely present in the environment today.
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 looking at hormone levels throughout the monthly cycle to help identify the best way and time to help.



Our customers are saying...
"I thought how fascinating if I could know more about my future. I should know. I should be aware for myself. For my children. If there's something that I could prevent for the future, or live my life in a different way, why not learn? Why not help myself? And be knowledgeable in that information for my health and well-being."

~ Susan M.

deCODE ME

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deCODE genetics
 the pioneers in gene discovery

deCODEme
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Susceptibility?

- From genetics to genomics
- The end of genetic determinism
- From 'biology is destiny' to knowing, managing and optimising biomedical futures
- From mutations – “the gene for” to SNPs for susceptibilities to common complex disorders, e.g. depression.
- Genome Wide Association Studies
 - The new logic, despite many flaws
 - (e.g. SNPs identified by GWAS account for less than 20% of familial heritability of breast cancer)
- From “Genes-R-Us to “G x E”
- Susceptibility as a form of life
 - With its own obligations and forms of responsibility

2007: The Year of GWA Studies?

Consistently replicated associations found for:

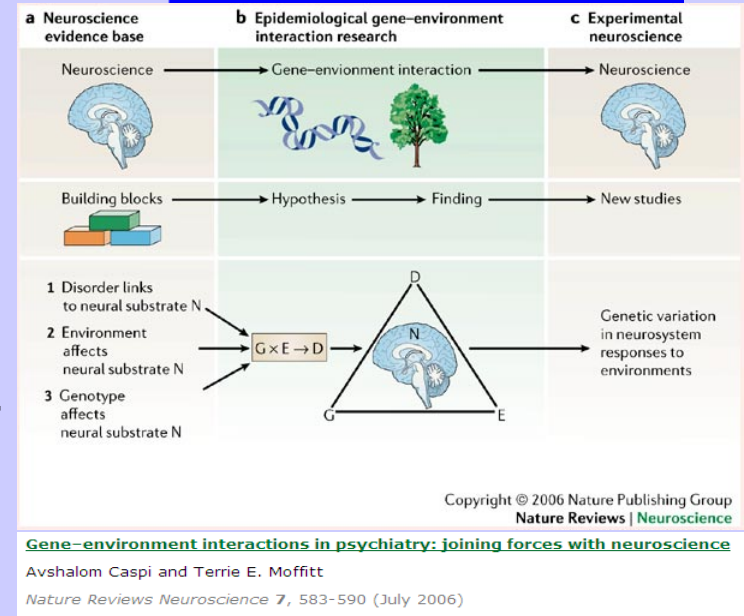
- 10 Jun 2007: Celiac disease
- 1 Jul 2007: Atrial fibrillation
- 8 Jul 2007: Colorectal cancer
- 15 Jul 2007: Gallstones
- 18 Jul 2007: Periodic limb movements in sleep
- 19 Jul 2007: HIV viral setpoint
- 26 Jul 2007: Childhood asthma
- 29 Jul 2007: Multiple sclerosis
- 1 Aug 2007: Amyotrophic Lateral Sclerosis
- 9 Aug 2007: Exfoliation glaucoma
- 2 Sep 2007: Height
- 5 Sep 2007: ??

A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants

Genome-Wide Association Analysis Identifies Loci for Type 2 Diabetes and Triglyceride Levels

Replication of Genome-Wide Association Signals in UK Samples Reveals Risk Loci for Type 2 Diabetes

Eleftheria Zeggini,^{1,2*} Michael N. Weedon,^{3,4*} Cecilia M. Lindgren,^{1,2*} Timothy M. Frayling,^{3,4*} Katherine S. Elliott,² Hanaango,^{3,4} Nicholas J. Timpton,^{3,4} John B. Perry,^{3,4}



Biomedical patenting – a matter of life or death?
Michael Crichton, New York Times, February 2007

- “YOU, or someone you love, may die because of a gene patent that should never have been granted in the first place. Sound far-fetched? Unfortunately, it’s only too real.”
- **“Gene patents are now used to halt research, prevent medical testing and keep vital information from you and your doctor. Gene patents slow the pace of medical advance on deadly diseases. And they raise costs exorbitantly: a test for breast cancer that could be done for \$1,000 now costs \$3,000.”**
- “Why? Because the holder of the gene patent can charge whatever he wants, and does. Couldn’t somebody make a cheaper test? Sure, but the patent holder blocks any competitor’s test. He owns the gene. Nobody else can test for it. In fact, you can’t even donate your own breast cancer gene to another scientist without permission. The gene may exist in your body, but it’s now private property.”
- Was Crichton right?

Personalised – from 'we' to 'me'

BCM View of Personalized Medicine

- Best service
- Best quality based on evidence
- Individually tailored
- Backed by best science
- Genetically specific
- Fully Integrated
- Delivers best care and value
- Patient involvement



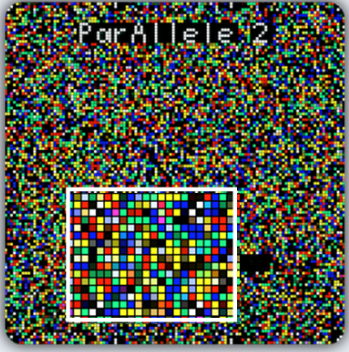
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BESTMINDS BESTMEDICINE™

Johnson promises personalised NHS

Deborah Summers and agencies
guardian.co.uk, Sunday March 2 2008
Article history · Contact us



Alan Johnson, the health secretary. Photograph: Kirsty Wigglesworth/PA

The days of a "one-size-fits-all" NHS are over, Alan Johnson claimed today.

The health secretary used Labour's spring conference in Birmingham to set out the government's promise of a more personal health service

The Age of Personalized Medicine

A Service of **PMC** Personalized Medicine Coalition
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Personalized Medicine

A new era of healthcare through:

Better medical outcomes.
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View of the Experts

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PMC Personalized Medicine Coalition

THE CASE FOR PERSONALIZED MEDICINE

We present a case for personalized medicine, shedding light on its demonstrated benefits and limitations, and outlining a realistic scenario for its evolution.

Perils of PPP

- Reliability: Risk assessments vary between providers
- Validity: Even if assessments are reliable, they have little or no clinical relevance
- Results (risk information) difficult to interpret without much additional knowledge (e.g. population risk)
- Can't take account of factors other than genetic
- Evidence suggests people overestimate certainty and underestimate uncertainty
- In most cases there is little preventive action available apart from sensible lifestyle choices
- Consumerization: individualisation of risk and security
- Responsibilization: May increase belief in individual 'responsibility' for a future that cannot fully be known or managed

1 Janssens, Gwinn and Bradley *et al.* (2008) A critical appraisal of the scientific basis of commercial genomic profiles used to assess health risks and personalize health interventions. *American Journal of Human Genetics* 82(3): 593–9.

Predictive: Screen and Intervene:

- “Earlier is (almost) always better...” – is it?
- Screening – who benefits?
 - Breast cancer?
 - Prostate cancer?
 - Psychopathy (children)?
 - Dementia (Mild Cognitive Impairment...)?

Published 24 September 2009, doi:10.1136/bmj.b3572

Cite this as: BMJ 2009;339:b3572

Editorials

Prostate specific antigen for detecting early prostate cancer

Evidence is inconclusive, so patient education and shared decision making are essential

The
Washington
Post

Article: Analysis: Mammograms Don't Cut Cancer Death Risk; Danish Researchers Find No Reliable Evidence in Major Studies to Support Medical Consensus

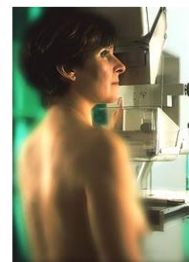
Article from: The Washington Post | Article date: October 19, 2001 | Author: Susan Okie |

Published online 19 October 2001 | Nature | doi:10.1038/news011025-5

News

Is screening for breast cancer with mammography justifiable?

Study questions whether mammography saves lives.



Mammography screening may not stand up to scrutiny. © SPL

Breast-cancer screening programmes may not save lives, according to a new examination of clinical trials. The controversial findings have led to calls for a re-evaluation of the routine monitoring procedure undergone by numerous women.

Mammography, X-ray breast imaging, is used in Europe and the United States to catch cancers early. "We've based a national screening programme on a set of results which do not stand up to scrutiny," says Richard Horton, editor of *The Lancet*, the medical journal that today publishes the peer-reviewed research.

In January 2000, Ole Olsen and Peter Gotzsche of the Nordic Cochrane Centre in Copenhagen, Denmark first questioned whether regular screening of middle-aged women reduces the overall cancer death rate¹. Their challenging re-analysis of large-scale mammography trials sparked public and medical controversy as some questioned the validity of the work.

Now Olsen and Gotzsche have confirmed their earlier conclusions with

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NHS screening programme takes centre stage in Brown fightback

• PM launches drive to avert 200,000 deaths a year
• New attempt to regain initiative from Tories

David Hencke, Westminster correspondent
The Guardian, Monday 7 January 2008
Article history



Gordon Brown talks to people who have found work through a job centre in central London. Photograph: Leftis Pitarakis/AP

The NHS is to launch a national screening programme to tackle some of Britain's biggest killer diseases, including heart attacks, strokes, diabetes and kidney failure, Gordon Brown will announce today. The programme, the first of its kind in the world, is expected to help prevent 200,000 deaths a year among the 6 million or more people who suffer from the targeted diseases, and heralds a switch in health priorities from curing illness to preventing it.

The announcement is intended to demonstrate the prime minister's commitment to making the "big

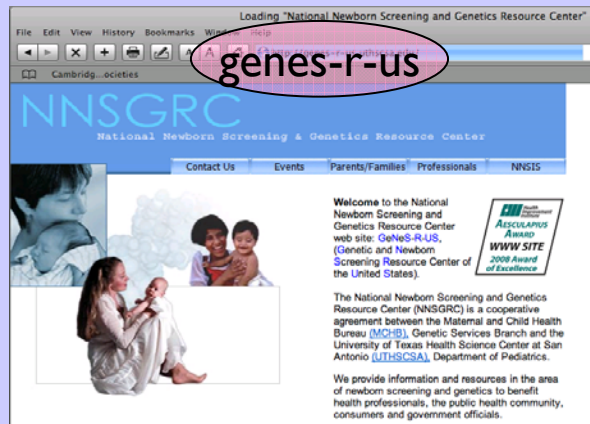
Saving lives
through screening



NHS Breast Screening Programme
Annual Review 2008

Cancer Screening Programmes

Screening – public value meets biovalue?



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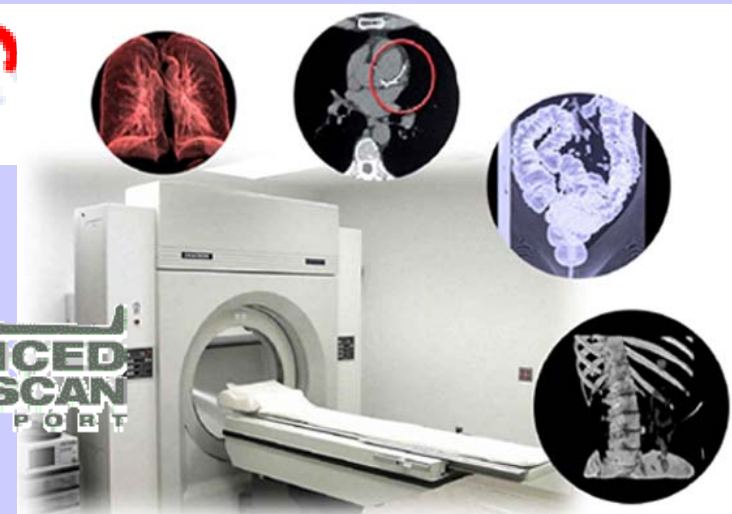
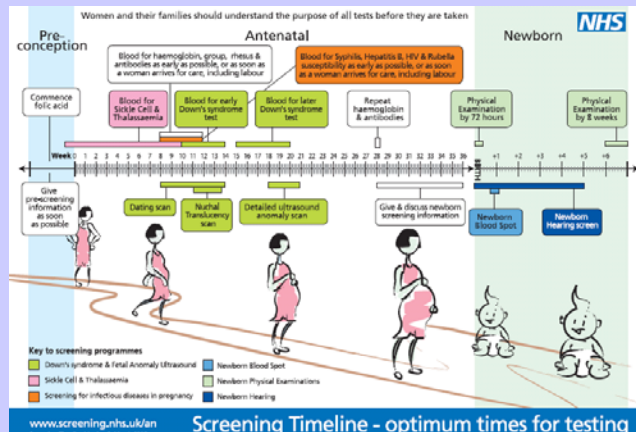
Schizophrenia Research 90 (2007) 130–146

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www.elsevier.com/locate/schres

Community screening for psychotic-like experiences and other putative antecedents of schizophrenia in children aged 9–12 years

Kristin R. Laurens^{a,*}, Sheilagh Hodgins^a, Barbara Maughan^b, Robin M. Murray^c,
Michael L. Rutter^b, Eric A. Taylor^d



Biomarkers in psychiatry

THE LANCET Neurology

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Review

The Lancet Neurology, Volume 7, Issue 8, Pages 704 - 714, August 2008
doi:10.1016/S1474-4422(08)70162-5 [Cite or Link Using DOI](#)

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Biomarkers for cognitive impairment and dementia in elderly people

Joshua A Sonnen MD ^a, Kathleen S Montine PhD ^a, Joseph F Quinn MD ^b, Jeffrey A Kaye MD ^b, John CS Breitner MD ^c, Thomas J Montine MD ^a 

Summary

The threat of a looming pandemic of dementia in elderly people highlights the compelling need for the development and validation of biomarkers that can be used to identify pre-clinical and prodromal stages of disease in addition to fully symptomatic dementia. Although predictive risk factors and correlative neuroimaging measures will have important roles in these efforts, this Review describes recent progress in the discovery, validation, and standardisation of molecular biomarkers—small molecules and

NEWS



p5 Abstinence all the way: New US family planning chief slams premarital sex.



p7 Vietnam's winner: Menno de Jong fights for local scientists' rights.



p10 Acid test: Many with painful cluster headaches are self-medicating with LSD.

Biomarkers trump behavior in mental illness diagnosis

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Here's how doctors decide which mental or neurological disorder their troubled patients suffer from: they ask questions like, "Are you hearing voices?" and "Do you feel like people are out to get you?"

Not all that different from how they used to do it about 100 years ago.

New techniques are set to radically change that approach—and perhaps define new categories within each disease—relying more on changes in physiology than in behavior.

"Now we're at the opposite end of the spectrum where we can take an unbiased look," says Stephen Clark, a psychiatric geneticist at the State University of New York in Syracuse.

Delusions, hallucinations, disorganized thinking and other psychotic symptoms can result from schizophrenia, Alzheimer disease, bipolar disorder, manic depression or dementia. Even diabetes and syphilis can induce forms of psychosis.

The answers to doctors' questions can help whittle down the categories somewhat, but it's far from a perfect science. Less than a third of individuals with these disorders respond to medications, most likely because they are being treated for the wrong one. The treatment for each

illness can be drastically different, so pinning down the diagnosis is crucial.

Scientists are increasingly turning to biomarkers—such as genes or proteins in tissues, blood and body fluids—to distinguish between symptomatically indistinct illnesses.

"This is the holy grail of research," says Karoly Mirnics, associate professor of medical psychiatry at Vanderbilt University. Using techniques that have only become available in the past few years, scientists are looking at the brain, serum and spinal fluid and asking which combinations of genes or proteins might be expressed differently in these disorders.

The field is still far from ready for the clinic, but holds great promise: markers could allow for earlier treatment and a better prognosis, ultimately enabling scientists to intervene even before the full-blown disease strikes.

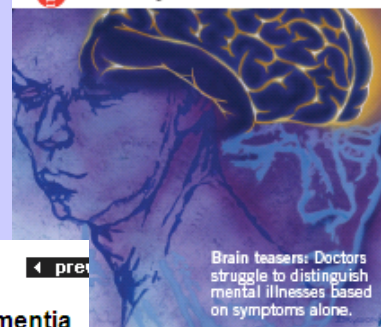
In one study published in November, researchers tested spinal fluid from patients at the first onset of schizophrenia. They found higher levels of a small protein derived from the protein VGF, known to be important for energy metabolism, and lower levels of transthyretin compared with

scientists are examining panels or 'pathways' of biomarkers. Even within the umbrella of a single disease as defined clinically, there may be several subtypes that could be distinguished at a molecular level.

For example, "It's very unlikely that a schizophrenic from one end of the spectrum to a schizophrenic on the other end of the spectrum will show the exact same biomarkers," says Mirnics.

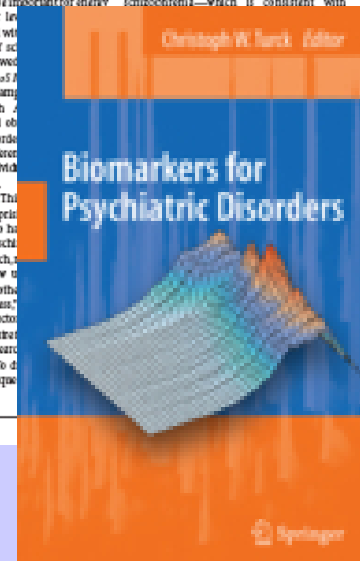
In a study published in August, Baker's group also found that treating schizophrenics who had had only one psychotic episode—but not more than one—corrected abnormal patterns in the expression of some metabolites, including glucose, lactate and acetate (PLoS Med. 3, e127), underscoring the importance of early intervention.

Biomarkers are also revealing unexpected insights into the biology of diseases. For example, using gene chips, one team found that some pathways involved in energy and metabolism are affected in individuals with schizophrenia—which is consistent with



Brain teasers: Doctors struggle to distinguish mental illnesses based on symptoms alone.

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


Biological Psychiatry

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Cerebrospinal Fluid Biomarkers in Parkinson's Disease with Dementia and Dementia with Lewy Bodies

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Mild Cognitive Impairment Aricept And Alzheimer's Disease

Treatment for MCI: Is the evidence sufficient?

Neurology - Volume 70, Issue 22 (May 2008) - Copyright © 2008 American Academy of Neurology - [About This Journal](#)

Editorials

Treatment for MCI: Is the evidence sufficient?

Paul S. Aisen, MD

From the Department of Neurology

Self-Administered Screening for Mild Cognitive Impairment: Initial Validation of a Computerized Test Battery

Jane B. Tortore, Ph.D.
Emory Hill, Ph.D.
Jo Anne Laboff, M.S.W.
Mary E. McGann, M.P.H., M.S.W.



SCREENS TEST BATTERY

The CANS-MCI

Screen's test battery is named the "Computer-Administered Neuropsychological Screen for Mild Cognitive Impairments"—or CANS-MCI for short.

Screen's Test Battery meets high statistical standards and is extremely valuable in both pre-diagnostic and post-diagnostic procedures.

70 DIAGNOSIS DemTect effective in screening for mild cognitive impairment and mild dementia

Kalish E, Kessler J, Calabrese P, et al. DemTect: a new, sensitive cognitive screening test to support the diagnosis of mild cognitive impairment and early dementia. *Int J Geriatr Psychiatry* 2004;19:135-43.

Does the DemTect reliably detect mild dementia and mild cognitive impairment in older adults?

METHODS

- Design: Prospective cohort study.
- Setting: Three centres in the UK; telephone not used.
- People: 121 people aged 65-92 years with possible mild to moderate Alzheimer's disease (NINCDS-ADRDA criteria). Clinical Dementia Rating scale (CDR) 1 or 2; 87 people aged 65-92 years with mild cognitive impairment (MCI) (MCI criteria); CDR 0.5-1.45 (average aged 65-92 years with no cognitive impairment (CDR 0). Control group was divided into 430 years and 430 years. Alzheimer's disease group was divided into Mild/Moderate (MMSE 24-27) and MCI (MMSE 21-23).
- Test: The DemTect includes five short, easy-to-administer tests that are sensitive for detecting dementia (word list, delayed word list, word fluency, sentence word fluency, digit span reversal).
- Diagnostic standard: Full clinical assessment plus CDR to assess dementia and MCI criteria to assess mild cognitive impairment.
- Outcome: Sensitivity and specificity of DemTect and MMSE in classifying people with mild to moderate Alzheimer's disease and mild cognitive impairment compared with full clinical assessment.

MAIN RESULTS

The standardized total DemTect score is independent of age and education and performed well when compared with MMSE. At a DemTect cut off score of 13, overall classification rate, sensitivity and specificity were high. (See table 1). Score of 13-18 represents appropriate cognitive power for age. Additional cut off scores were

useful for predicting mild cognitive impairment and dementia (mild cognitive impairment: 9-12 points; dementia: 0-8 points; total classification rate 85.4%).

CONCLUSIONS

The DemTect is a useful scale for identifying mild cognitive impairment and early dementia in older adults. It is easy to administer and accepted well by participants.

Commentary

Clinicians are increasingly recognizing the importance of detecting the presence of mild cognitive impairment (MCI) in their patients. MCI is associated with a five to tenfold increased risk of developing dementia compared with cognitively healthy individuals. Diagnosis of MCI requires both a clinical interview and detailed psychometric testing. In order to identify individuals who require further assessment, clinicians need a tool to screen for MCI using sensitive screening tools.

The DemTect was designed for this purpose. Kalish et al report the development and clinical effectiveness of the English version of the test. The strengths of the DemTect include (1) short administration and scoring time (5-10 minutes); (2) assessment of multiple cognitive areas sensitive to MCI and dementia, including immediate and delayed memory, number sequencing, semantic fluency, and working memory; (3) high sensitivity in detecting MCI and early Alzheimer's disease, as identified with the Clinical Dementia Rating Scale; and (4) a large range of scores in the mild impairment range, allowing detection of subtle changes over time.

The DemTect would appear a very good tool for the repeated purpose of screening for MCI, and seems to be superior to the Mini-Mental State Examination (MMSE) for this purpose—test is individuals with MCI obtain scores below the normal range on the DemTect, but generally within the normal range on the MMSE. On the other hand, the DemTect does not seem to be sufficient for the diagnosis of MCI, because diagnosis requires psychometric evidence of both a mild memory impairment (which DemTect can provide) and three general cognitive functions (for example, naming, visual construction, attention, which DemTect does not provide).

For the DemTect to be used clinically, the actual test materials and the scoring key must be available to the clinician. The authors state that the

Table Sensitivity and specificity of DemTect and MMSE compared

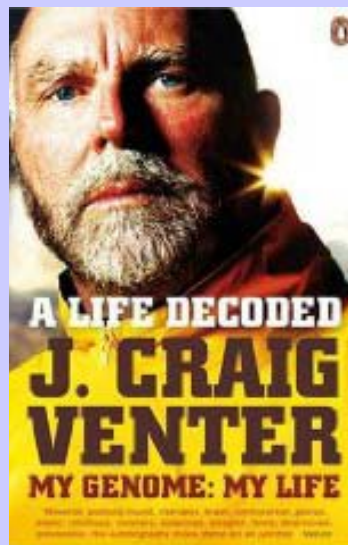
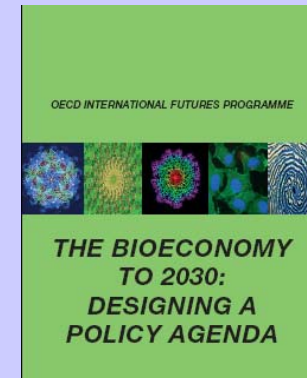
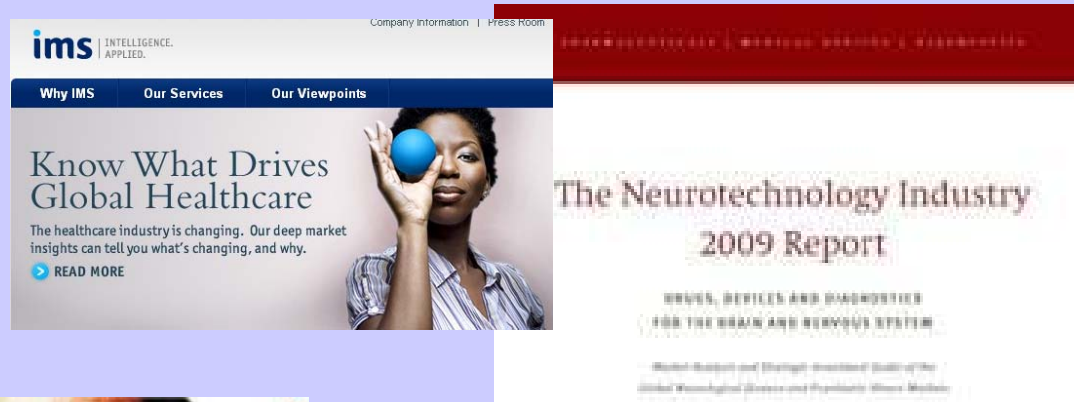
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Screening for Dementia in Primary Care Summary of the Evidence

Maria Bouillon, M.D., M.P.H., Britt Peterson, M.D., M.P.H., Laura Hanson, M.D., M.P.H., Russell Harris, M.D., M.P.H., Kathleen L. Lohr, Ph.D.

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The economic drivers of biomedical technology

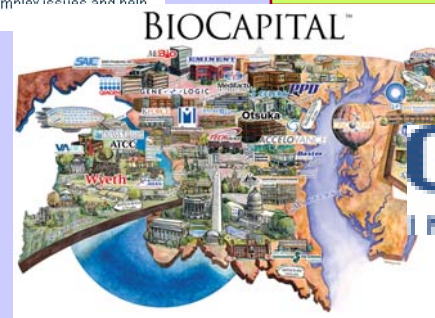


OECD (2006) 'the bioeconomy' is "that part of economic activities "which captures the latent value in biological processes and renewable bioresources to produce improved health and sustainable growth and development"

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Thank you for your attention!



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