

PERSONALIZED HEALTH CARE: THE GENETIC CHALLENGE

*Preventative strategies for
addressing chronic diseases...*

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Health Law Institute



Stem Cell
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A H F M R

ALBERTA HERITAGE FOUNDATION
FOR MEDICAL RESEARCH

Selection of Issues (all need work!)

DTC Testing and Popular Culture

Impact on Health Systems

Health Benefits?

Patent Problems

Confidentiality/Privacy

Consent Challenges

What price personal genome exploration?

Control of direct-to-consumer genetic testing

NEWS

Accessible genomes move closer

NEW YORK Rarely do research subjects attend scientific meetings. Yet at the inaugural Cold Spring Harbor Personal Genomes Harbor meeting this month on Long Island in New York, the Nobel-prizewinning biologist James Watson sat in the front row as other researchers dissected his genetic vulnerabilities via a PowerPoint presentation. So far, Watson says, it has not been a particularly profound experience. "I haven't really learned anything, except that I'm lactose intolerant!"

Other than Watson, only three people have had their genomes sequenced in full, genomes sequencing efforts but other sequencing efforts are under way, including the 1,000 Genomes Project and Personal Genomes Project. And researchers may soon be able to start linking this flood of personal genomic data to the

Exciting as it is, the prospect of multiple genomes per hour poses daunting data challenges, says Elaine Mardis, co-director of the Genome Sequencing Center at Washington University in St. Louis. She is leading a project to develop tools that will become more powerful as new projects come on line. "We need to do that with the tools we have now, and we need to develop tools to analyze the data that will come out of these projects," she says.

Radoje Drmanac, et al., "Human Genome Sequencing Using Unchained Base Reads on Self-Assembling DNA Nanoarrays" Science (2010) - \$4500 sequences!

The human genome project has spawned a new commercial field, molecular diagnostics. Some companies charge as much as US\$350,000 to scan a person's entire genome to forecast disease risk. Other companies claim to be able to diagnose and determine the most effective treatment for

of companies marketing genetic tests without acceptable proof of clinical reliability.

Last week, the FDA made a positive move. A warning letter sent to the Laboratory Corporation of America, the nation's second-largest clinical laboratory company, states the company's InViaSure test is illegally marketed and poses a potential public-health risk. Introduced in June, the test analyzes six proteins in blood to assess whether or not a woman has ovarian cancer. Because the test was

www.nature.com/nature

My genome. So what?

Research is needed into the way individuals use information, and into p

Human genome research anticipated, the speed of disease has quickened, and fed back to them in electronic groups reveal individual genomic page 53), and of a Han Chinese — each — a fraction of that of the editions.

The age of personal genomes

Basic science, targeted therapies, human history...

EDITORIAL

1 Mygenome.

By AMY HARMON

In 2001 Collins and Mansoura declared that the "the most critical measure of the success of the HGP will be determined by the answer to this question: To what extent did the scientific and medical advances derived from the HGP reduce the burden of disease for all people?"

As the first conference on personal genomics in New York, this month at Cold Spring Harbor Laboratory, the extent was a little premature. After all, only four people's genomes have so far been fully sequenced and assembled, and it's still quite difficult to interpret the genetic variation found in them (see page 1014). But the participants soon began to realize that, in one sense, the meeting was overdue. Increasingly, private companies are offering personal genome scans. And genetic tests for sale — and consumers are buying them. Meanwhile, some scientists earlier this week made public parts of their genetic and medical data through the Personal Genome Project,

October 20, 2008

The DNA Age

Taking a Peek at the Experts' Genetic Secrets

BOSTON —

Vol 45

Does so much genetic information pose a major health risk?

...which his grand

...have a gene variant that raises his risk of

...to greater than 50 percent?

...he may have

...Monday, they may

...ers to these and other questions — and, if all goes according to

...ing, the public desperately wants help in making such a visit a public Web site, www.personalgenomes.org. The three

Part of the problem is the information overload provided by the

Internet. Consumers can point their browsers to a slew of content,

which can come from both trusted sources and charlatans. And in

the age of e-mail whisper campaigns, lies

can proliferate, opinions can replace facts,

and experts are no longer trusted to know the truth.

"Scientists cannot put the genie back in the bottle."

PRINTER-FRIENDLY FORMAT
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NOW PLAYING

training to be an astronaut,

...a gene variant that raises his risk of

...a high risk of breast cancer, which

...and, if all goes according to

...the three

The Future?

Oberg touts 'right to know' in promoting DNA test firm

By Keith Gerein, Edmonton Journal June 3, 2010

who led the Human Genome Project... affordable reality with... al DNA coding... the scientist... dicted.

<http://www.telegraph.co.uk/decade.htm>

June 12, 2010

NEW

A Decade Later, Genetic Map Yields Few New Cures

By [NICHOLAS WADE](#)

Ten years after President Bill Clinton announced that the field of personalized medicine has yet to see any large part of the promised benefit.

Gene Test for Dosage of Warfarin Is Rebuffed

By [ANDREW POLLACK](#)
Published: May 4, 2009

In a setback for the fledgling field of personalized medicine, [Medicare](#) has decided not to pay for genetic tests intended to help doctors determine the best dose of the blood thinner warfarin for a particular patient.

World still waiting for genome DNA map to unlock secrets

By Richard Gray, Daily Telegraph June 20, 2010

Canadian DTC Survey

(Ries, Hyde-Lay, Caulfield, (Public Health Genomics, 2010)).

Table 3: Willingness to Pay for Genetic Tests

Willingness to Pay for Genetic Testing	Manageable Disease	Serious, Unpreventable Disease	Determining Healthy Foods	Psychiatric Condition	Baldness (men only)	Risk of Gaining Weight
nothing	36.9	48.3	54.5	50.1	83.9	72.5
\$1-\$499	36.6	32.2	32.9	30.5	11.8	19.8
\$500-\$1999	17.5	12.6	8.7	12.5	2.5	5.2
\$2000+	9.0	7.0	3.9	6.9	1.8	2.5
Public Health System Should Pay*						
Disagree	21.9	37.0	38.6			
Neutral	16.5	18.9	20.0			
Agree	61.7	44.1	41.1			

*Respondents were only asked about publicly insured testing for the three tests indicated. The percentages for disagree and agree include responses of 'disagree/agree' and 'strongly disagree/agree'.

Canadian DTC Survey

(Ries, Hyde-Lay, Caulfield, (Public Health Genomics, 2010)).

Table 4: Factors that Influence Interest in Genetic Testing

	<i>No effect</i>		<i>Strong effect</i>		
	1	2	3	4	5
Availability of treatment	16.1	5	17.7	19.6	41.6
Curiosity	41	14	21.6	11	12.5
Reproductive decisions	44.4	7	17.7	12.4	18.5
Fear of discrimination	32.9	6.4	15.4	12.9	32.4
Healthy lifestyle choices	24.3	7.5	22.1	22.3	23.8

Reported as percentage of all respondents.

- Across all test categories, few respondents expressed willingness to pay more than \$500 out of their own pocket.
- Curiosity about genetic risk had only a modest impact on consumer interest.

Science's wild ride

Modify risk?

The New York Times

On the anniversary of genome
time to take stock of promise and adva

THE SUNDAY
SPECIAL

BY JAMES P. EVANS

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Commentary

Direct-to-consumers
David Magnus*,

Addresses: *Stanford Center for
& Policy, Institute for Genome

g. "There is absolutely no question," he said, "that
for the whole hope of personalized medicine, the news has
been just about as bleak as it could be."

But any direct payoff from such knowledge is years away. The idea that you will benefit in any tangible manner from knowing whether you are at an increased or decreased risk of, say, heart disease, is a fool's hope — in spite

2010

nature

OPINION

Has the revolution arrived?

Looking back over the past decade of human genomics, **Francis Collins** finds five key lessons for the future of personalized medicine — for technology, policy, partnerships and pharmacogenomics.

Third, the success of personalized medicine will depend on continued accurate identification of genetic and environmental risk factors, and the ability to utilize this information in the real world to influence health behaviours and achieve better outcomes. This will require

colon cancer, and lung cancer. Confronted with the reality of his own genetic data, he arrives at that crucial “teachable moment” when a lifelong change in health-related behavior, focused on reducing specific risks, is possible. And there is much to offer. By 2010,

Medicine

1999

Special Article

SHATTUCK LECTURE — MEDICAL AND SOCIETAL CONSEQUENCES
OF THE HUMAN GENOME PROJECT

FRANCIS S. COLLINS, M.D., PH.D.

Invention

What Your Gene Test Can Tell You

What Gene can be dis

40 genetic profiles of
8 major groups bel

IQ

23andMe



mygene profile

www.mygeneprofile.com/talent



Navigenics™

the autoimmune disorder Sjögren's
syndrome, which affects up to
8 million Americans

50% to 30% chance of
getting galactosea

50% lifetime chance
of getting colorectal
cancer

50% chance of getting the
bowel disease called Crohn's

20% to 70% chance of mental
dysfunction when taking certain
antidepressants

Health Behaviour Change



Genes and Behaviour

The Behavioral Response

“Any risk communication expert would laugh at the suggestion [that genetic risk information would motivate behaviour change] ... We have long known from other areas of research that the communication of risk is necessary but rarely sufficient. And even if people do change, they all relapse.”

Colleen McBride, Chief and Senior Investigator for the Social and Behavior Research Group at the National Human Genome Research Institute.

Saskia C. Sanderson,² and Kimberly A.


The few studies conducted to date based on single-gene probabilities has little impact—on or negative—on emotions, cognitions, or behavior. The difficulty of health behavior information may be less likely to achieve behaviour change than...
Weinman. 2006. Self-regulation and the behaviour research. S...
...information.” Marteau, T. M., and J. ...
...62: p.1360-1368.

Motivated Users?

Characteristics of users of online personalized genomic risk assessment: Implications for physician-patient interactions

Eric B. Larson, MD, MEd¹, Sharon Hensley Alford, PhD², Robert J. Reid, MD, PhD³,
C. Lee Jackson, PhD⁴, and Lawrence C. Brody, PhD⁴

tested. **Conclusions:** Individuals who present to health care providers with online genetics information may be motivated to take steps to change their lifestyle. These motives might be leveraged by health care providers to promote positive health outcomes. *Genet Med* 2009;11(8):582–587.



**Don't smoke. Maintain a healthy weight.
Exercise.
Eat fruits and vegetables.**

Most Heart Patients Skimp on Exercise After Rehab

A year later, only 37 percent were doing cardio exercises 3 times a week, study finds

-- Robert Preidt

Fetalism

ders

Dr. Joel Hirschhorn, a genetics and obesity researcher at Children's Hospital Boston, said people should not interpret the study to mean, "I don't have this gene variant so I don't need to be physically active."

Fighting the fat gene takes 3-4 hours a day

Or you can always live like the Amish, new research shows

Ap Associated Press

updated 2:14 p.m. MT, Mon., Sept. 8, 2008

Slide show

Maybe you CAN blame being fat on your genes. But there's a way to overcome that family history — just get three to four hours of moderate activity a day.

Sound pretty daunting?

Not for the Amish of Lancaster County, Pa., who were the focus of a new study on a common genetic variation that makes people more likely to gain weight. It turns out the variant's effects



smoking or change of diets."

Reduce Healthy Behaviour?

Impact on Health Systems

MAKES SIGH
DAACCTGG COPY
RIGHT TGAGGCAGGA
GTTG MOLECULAR ST
CACTGAGCCA CORRECT
AMYLOID TTTGGATCT
RISK INFORMATION AGT
G TRAIT'S REMEMBER
GA GENERATION AGGAG
SEARCH PEER CAGGA
GAGA REPEAT AAGA
ACCTCCAACA HERSELF
CCAACA AGAGCAAGGC
DISORDER PATIENCE
GTTTA SIGNIFICANT
WISHES OTHER TA
TGCTCATCTG TANGLES
GCATCT FUTURE

Impact on healthcare costs?

74% report they would use it to gain disease knowledge.

34% consider the information to be a medical diagnosis.

78% would ask their physician for help interpreting test results.

51% thought professional responsibility.

The American Journal of Bioethics, 9(6-7): 3-10, 2009
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DOI: 10.1080/15265160902928209

BMC Medical Ethics


BioMed Central

Research article

Open Access

Technology assessment and resource allocation for predictive genetic testing: A study of the perspectives of Canadian genetic health care providers

Alethea Adair^{†1}, Robyn Hyde-Lay^{*†1}, Edna Einsiedel² and Timothy Caulfield¹

ELSI ISSUES?

Commercialization

Marketing

Patents

Privacy

Education

Anxiety

Discrimination

Research priorities

Stigmatization

Genetisization

Hype/Public Trust

Conclusions

- Don't oversell (communication issues)
- Scientific progress is uncertain and iterative
 - Translation research
 - Behaviour change?
 - Fatalistic behaviour?
 - Increase healthcare costs?

Thank you!

• Amy Zarzeczny, Robyn Hyde-Lay, Ubaka Ogbogu, Nola Ries, Tania Bubela, Jim Evans, Amy McGuire, Wylie Burke, Therese Marteau, Eric Meslin and the HLI research team.



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