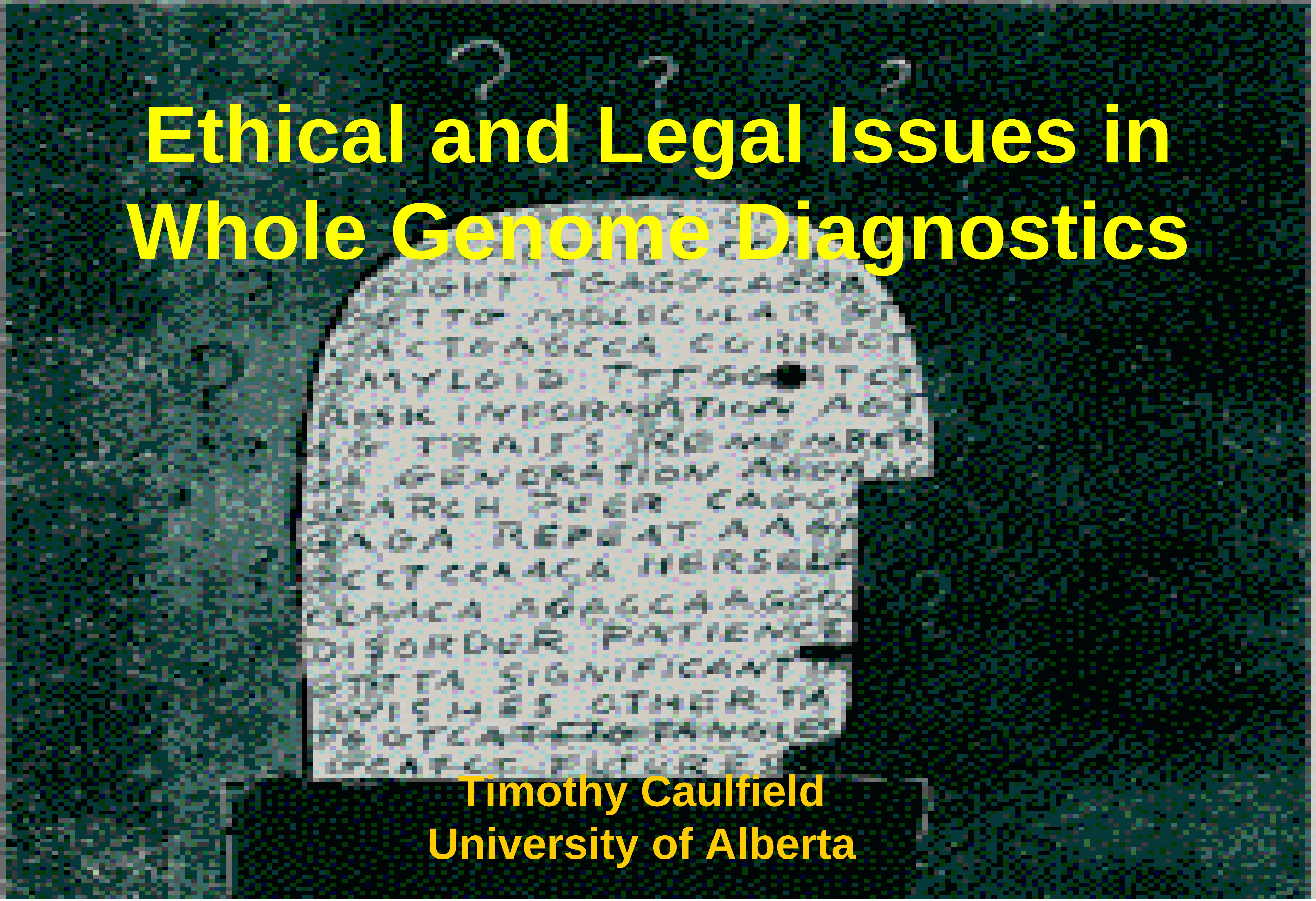
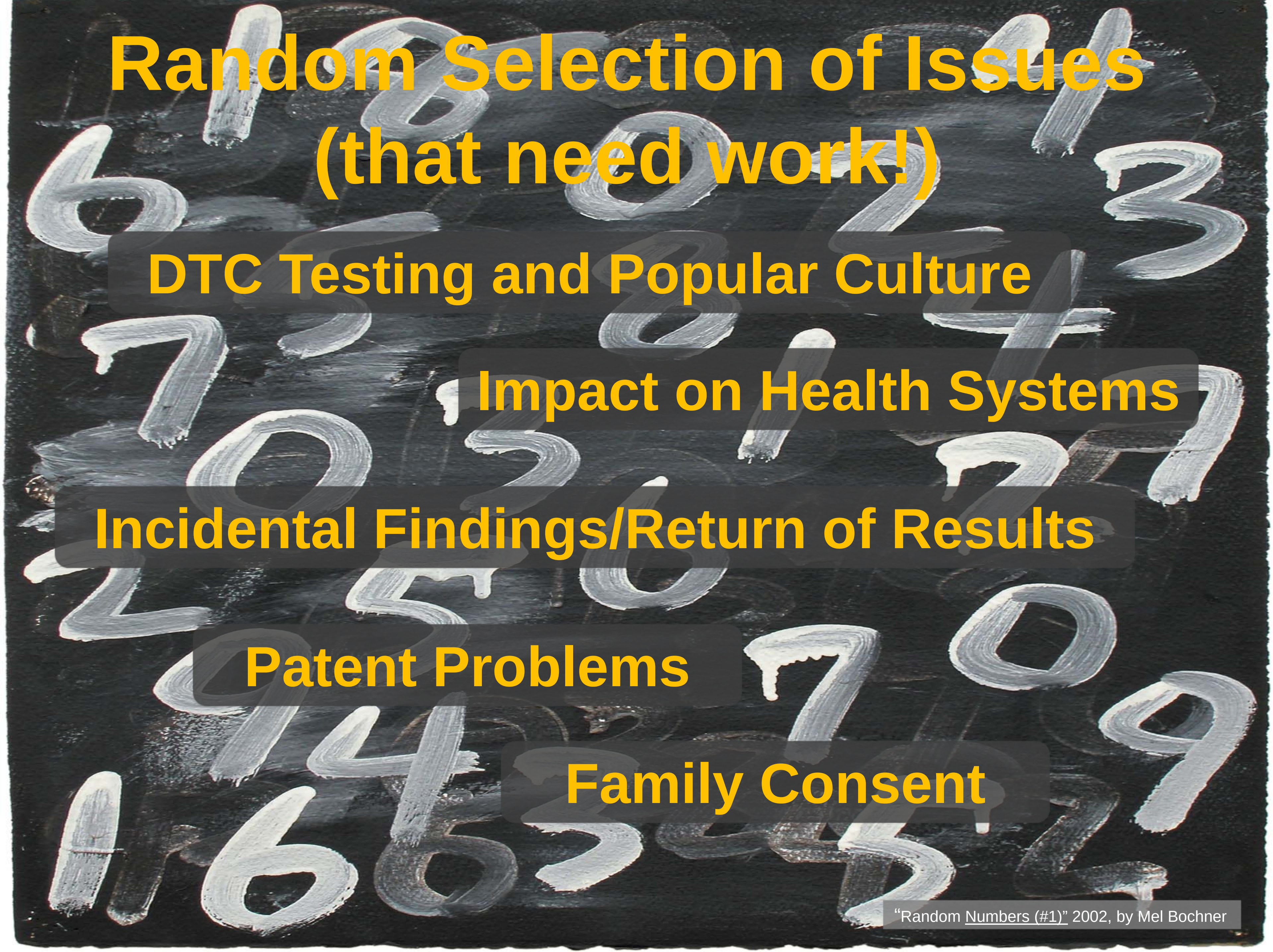


Ethical and Legal Issues in Whole Genome Diagnostics

Timothy Caulfield
University of Alberta

Health Law Institute
ge3ls ALBERTA
Stem Cell Network Réseau de cellules souches
AHFMR
ALBERTA HERITAGE FOUNDATION FOR MEDICAL RESEARCH



Random Selection of Issues (that need work!)

DTC Testing and Popular Culture

Impact on Health Systems

Incidental Findings/Return of Results

Patent Problems

Family Consent

“Random Numbers (#1)” 2002, by Mel Bochner

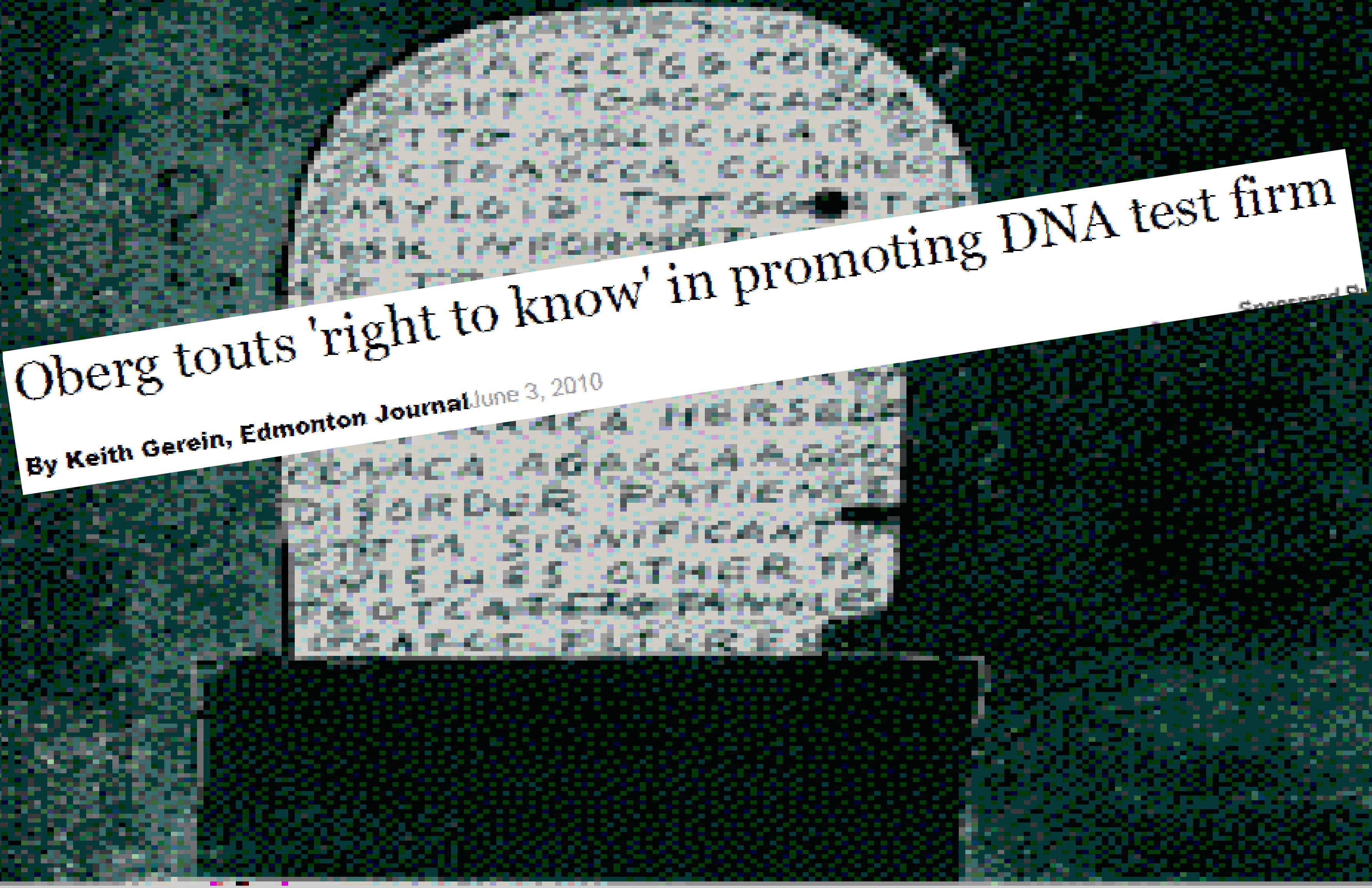
Massive challenge.

Few NEW issues.

(Just a bigger mountain)

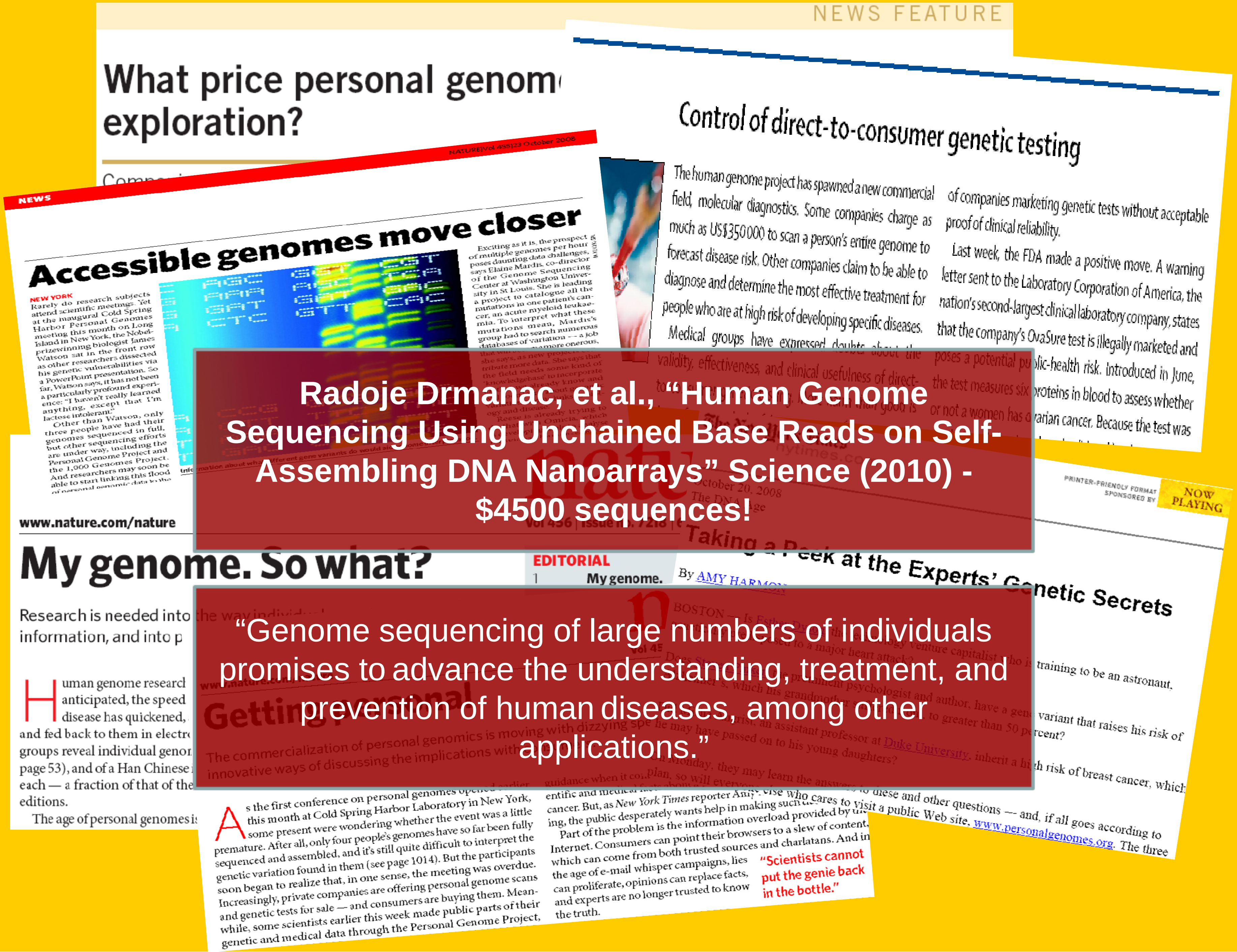
Few answers.

The DTC Debate



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

By Keith Gerein, Edmonton Journal June 3, 2010



NEWS FEATURE

What price personal genom
exploration?

NATURE|Vol 455|23 October 2008

NEWS

Accessible genomes move closer

NEW YORK Rarely do research subjects attend scientific meetings. Yet at the inaugural Cold Spring Harbor Personal Genomes 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, but other sequencing efforts are under way, including the Personal Genome Project and the 1,000 Genomes Project. And researchers may soon be able to start linking this flood

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 catalogue all the mutations in one patient's cancer, an acute myeloid leukaemia. To interpret what these mutations mean, Mardis's group had to search numerous databases of variation — a job

M. KULYK/SPL

Control of direct-to-consumer genetic testing

The human genome project has spawned a new commercial field, molecular diagnostics. Some companies charge as much as US$350000 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 people who are at high risk of developing specific diseases. Medical groups have expressed doubts about the validity, effectiveness, and clinical usefulness of direct-

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 that the company's OvaSure test is illegally marketed and poses a potential public-health risk. Introduced in June, the test measures six proteins in blood to assess whether or not a women has ovarian cancer. Because the test was

www.nature.com/nature

My genome. So what?

Research is needed into
information, and into p

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

The age of personal genomes i

EDITORIAL

My genome.

Getting personal

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 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,

cancer. But, as *New York Times* reporter Amy
ing, the public desperately wants help in making such

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."

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NOW PLAYING

Taking a Peek at the Experts' Genetic Secrets

By AMY HARMON

training to be an astronaut,

variant that raises his risk of
rcent?

h risk of breast cancer, which

who cares to visit a public Web site, www.personalgenomes.org. The three

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

"Genome sequencing of large numbers of individuals promises to advance the understanding, treatment, and prevention of human diseases, among other applications."

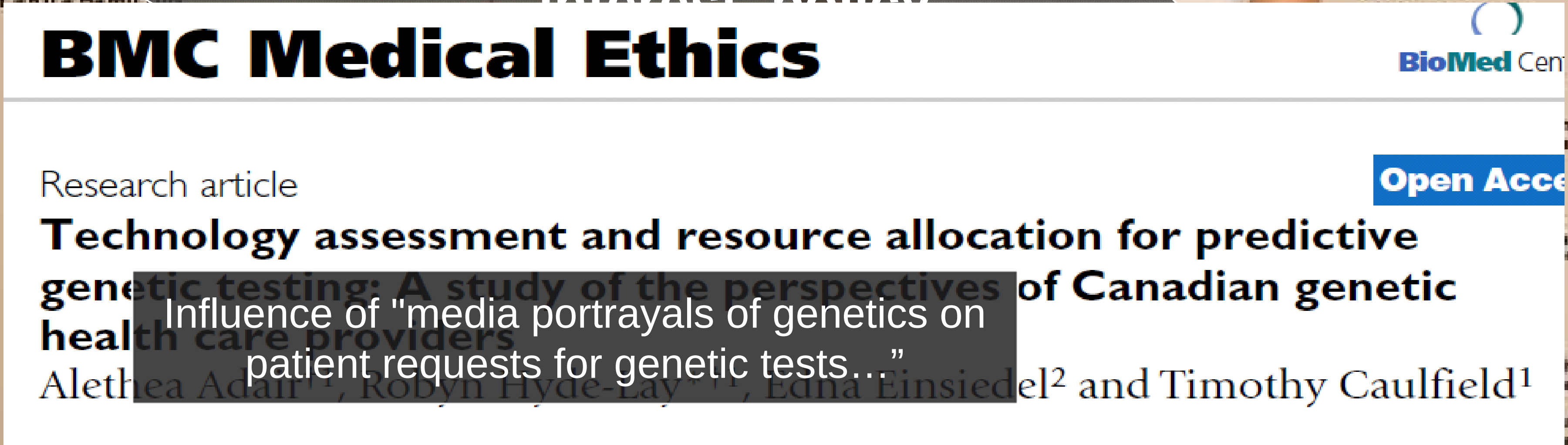
Invention Of the Year

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

BY ANITA HAMILTON

What Your Gene Test Can Tell You

Simplified view of role and place of genetics in our lives? Will this drive interest, policy



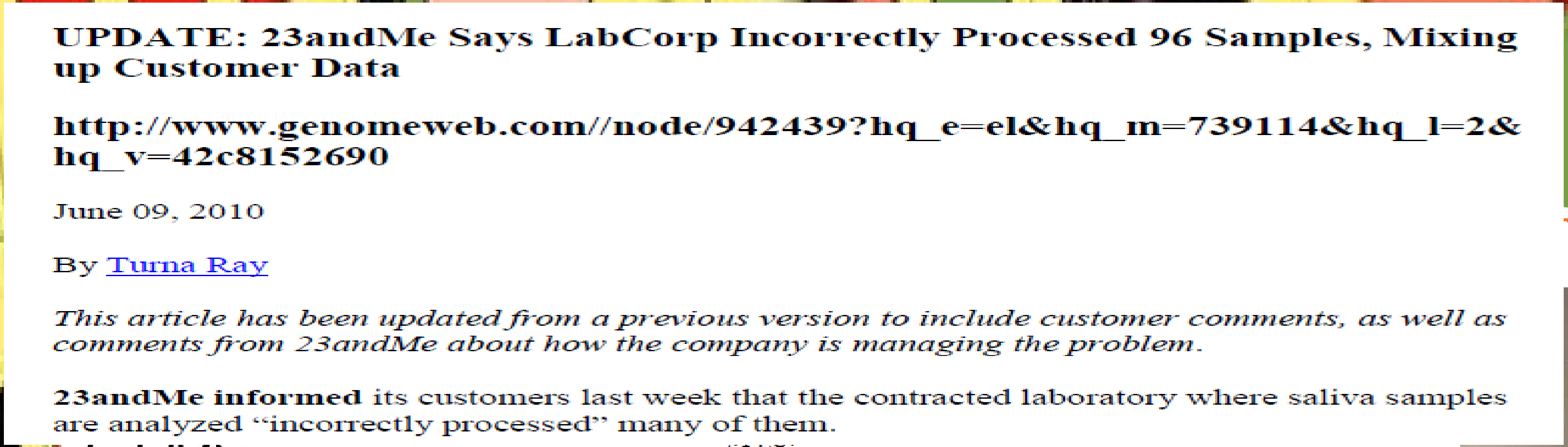
BMC Medical Ethics

Research article

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 Einsiedel[2] and Timothy Caulfield[1]

Influence of "media portrayals of genetics on patient requests for genetic tests…"



UPDATE: 23andMe Says LabCorp Incorrectly Processed 96 Samples, Mixing up Customer Data

http://www.genomeweb.com//node/942439?hq_e=el&hq_m=739114&hq_l=2&hq_v=42c8152690

June 09, 2010

By Turna Ray

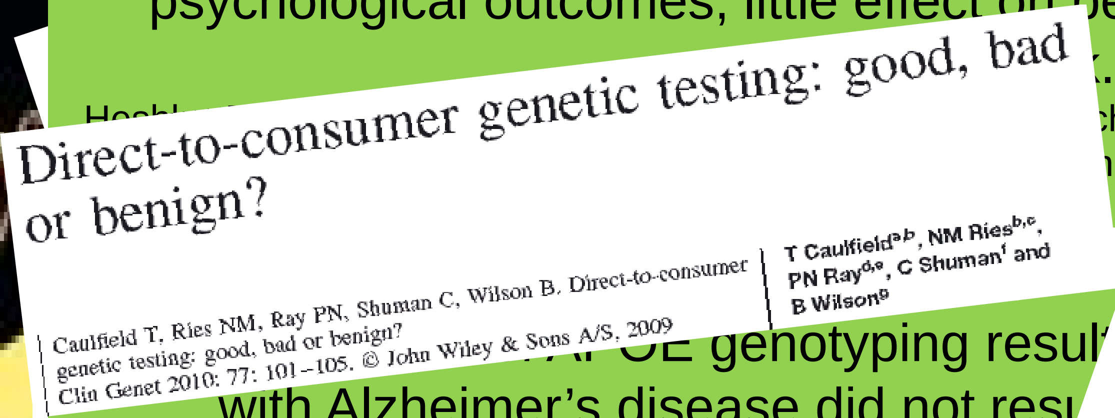
This article has been updated from a previous version to include customer comments, as well as comments from 23andMe about how the company is managing the problem.

23andMe informed its customers last week that the contracted laboratory where saliva samples are analyzed "incorrectly processed" many of them.

between the drugstore and Pathway Genomics , a San Diego company that markets at-home

“Overall, predispositional genetic testing has nc ificant impact on psychological outcomes, little effect on behavi change k.”

Hesh cholo ne, 2



Direct-to-consumer genetic testing: good, bad or benign?

Caulfield T, Ries NM, Ray PN, Shuman C, Wilson B. Direct-to-consumer genetic testing: good, bad or benign? Clin Genet 2010: 77: 101–105. © John Wiley & Sons A/S, 2009

T Caulfield[a,b], NM Ries[b,c], PN Ray[d,e], C Shuman[f] and B Wilson[g]

Analysis of Privacy Policies and Practices of Direct-to-Consumer Genetic Testing Companies: Private Sector Databanks and Privacy Protection Norms

Report funded by the Office of the Privacy Commissioner of Canada

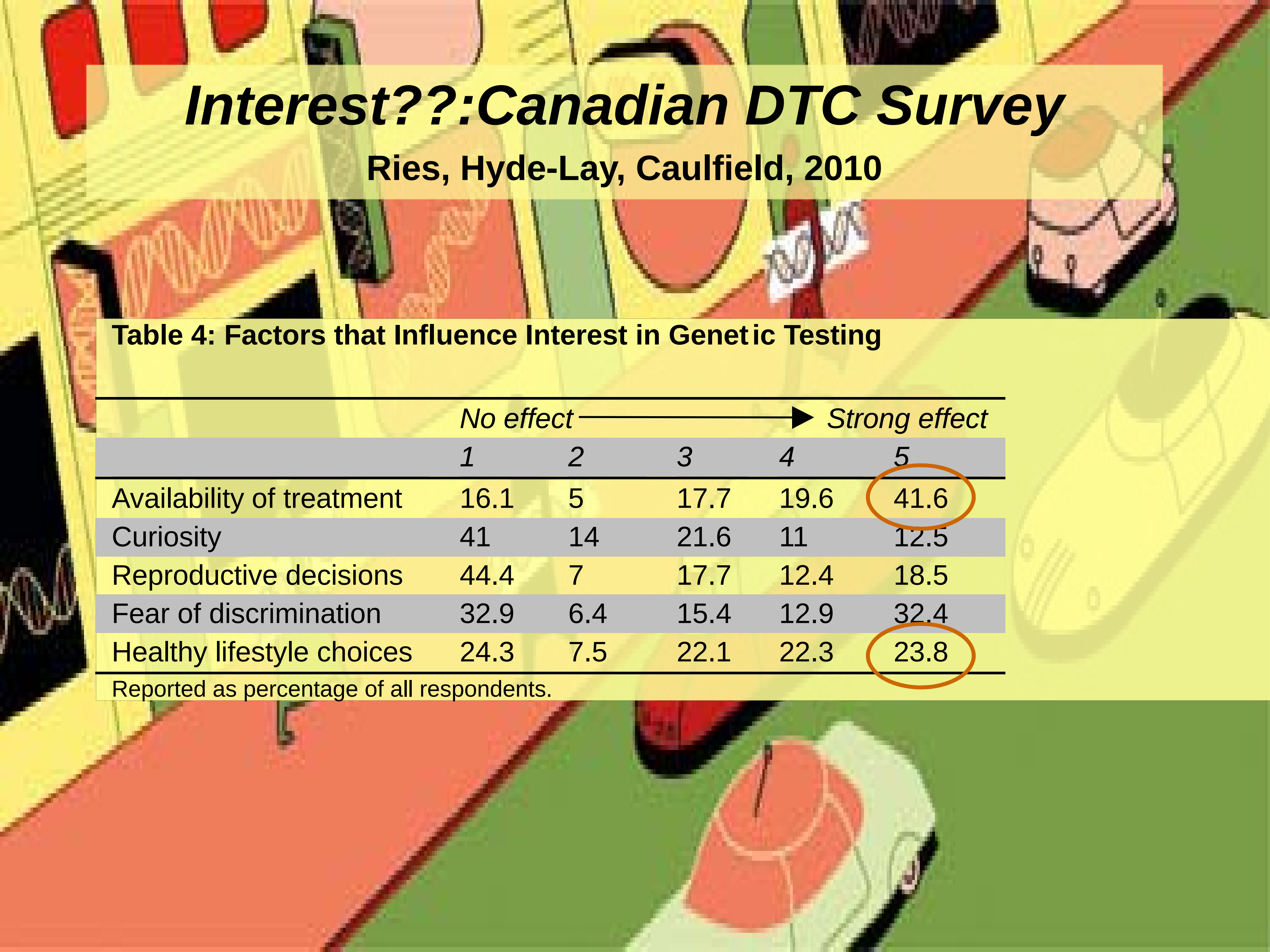
March 2010

Health Law Institute, University of Alberta

APOE genotyping result with Alzheimer’s disease did not resu psychological r

Green et al., NEJM, 2009

personal genetic health disposition results for more than 70 health conditions, including pharmacogenetics (prescription medication response), propensity for complex disease,



Interest??:Canadian DTC Survey

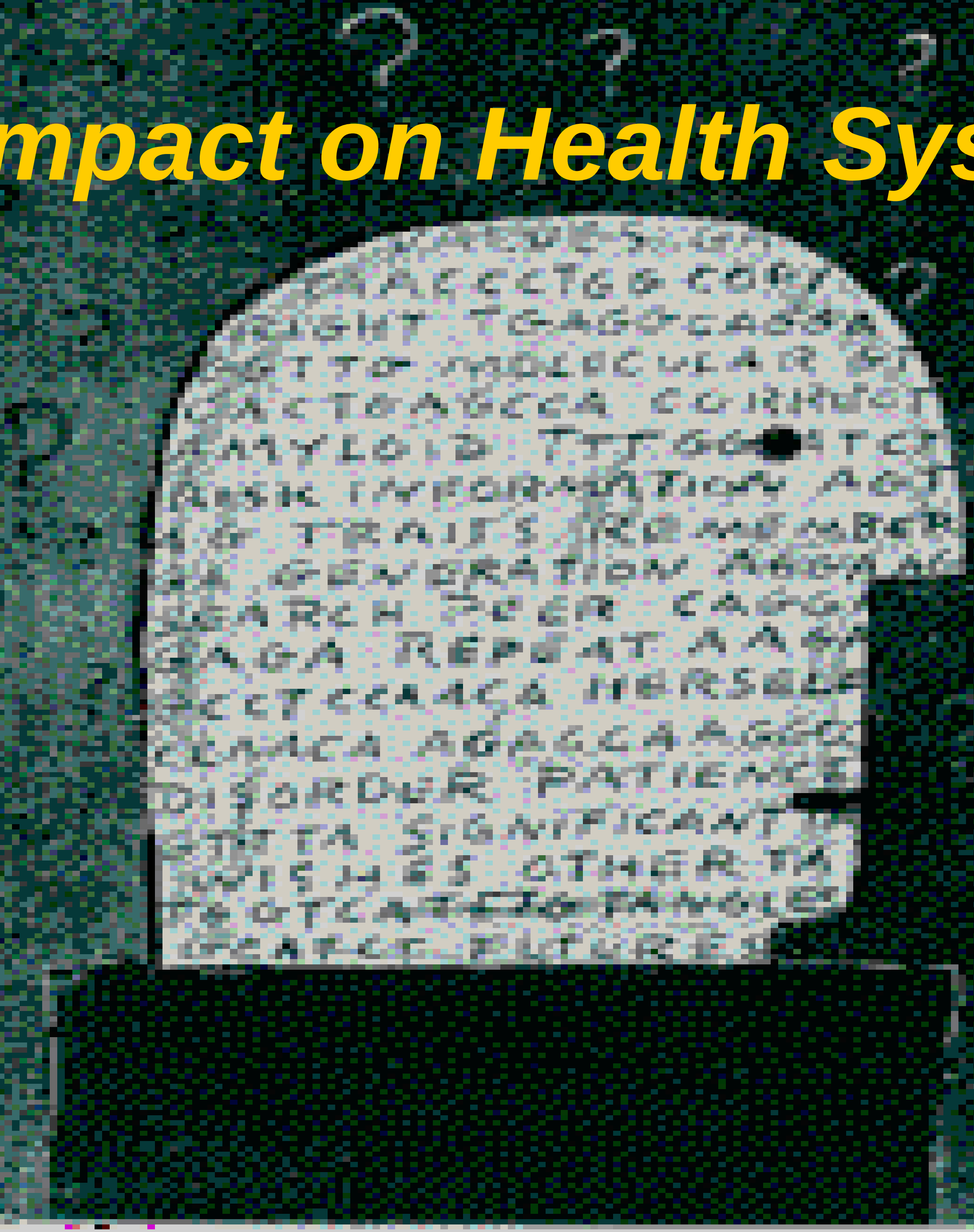
Ries, Hyde-Lay, Caulfield, 2010

Table 4: Factors that Influence Interest in Genetic Testing

	No effect →				*Strong effect*
	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.

Impact on Health Systems



Impact on healthcare costs?

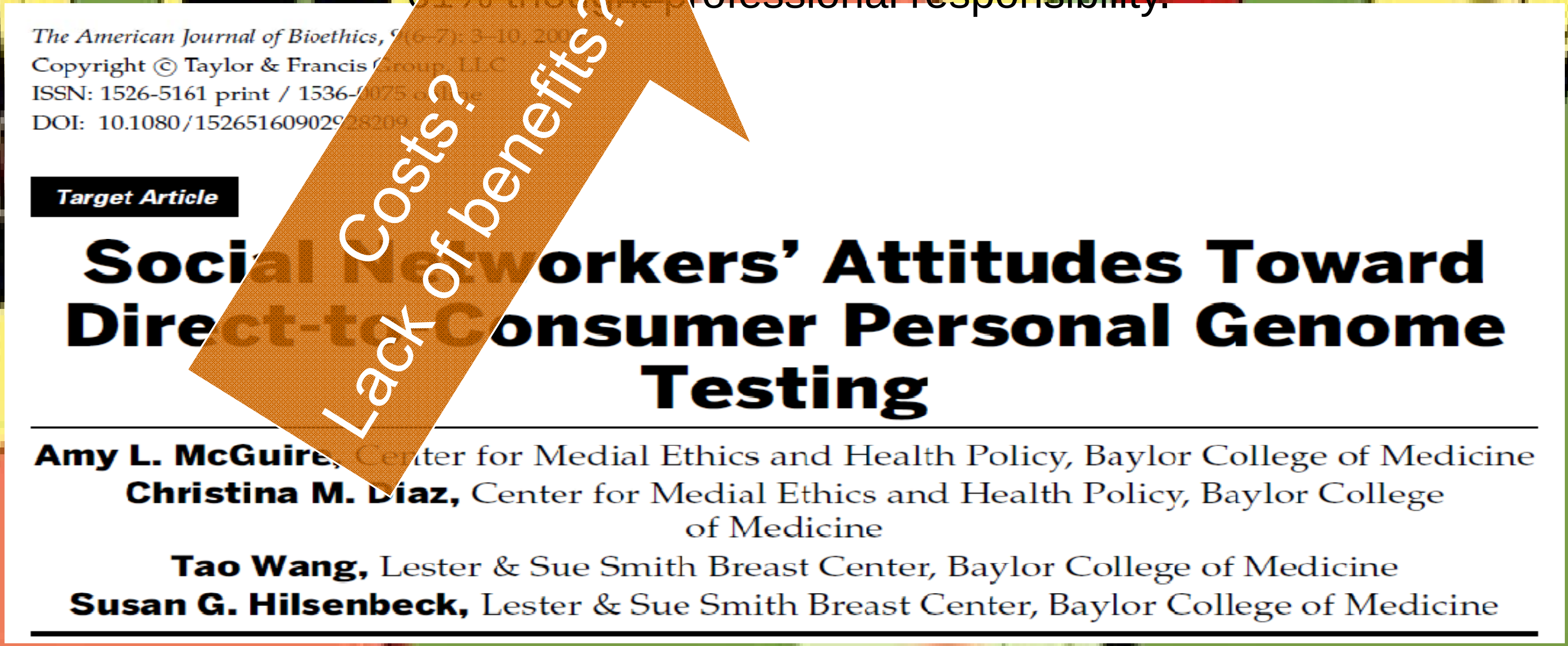
74% report they would use it to gain disease knowledge.

34% consider the information to be a medical diagnosis.

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

61% thought professional responsibility.

Costs?
Lack of benefits?



The American Journal of Bioethics, 9(6–7): 3–10, 200

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ISSN: 1526-5161 print / 1536-0075 online

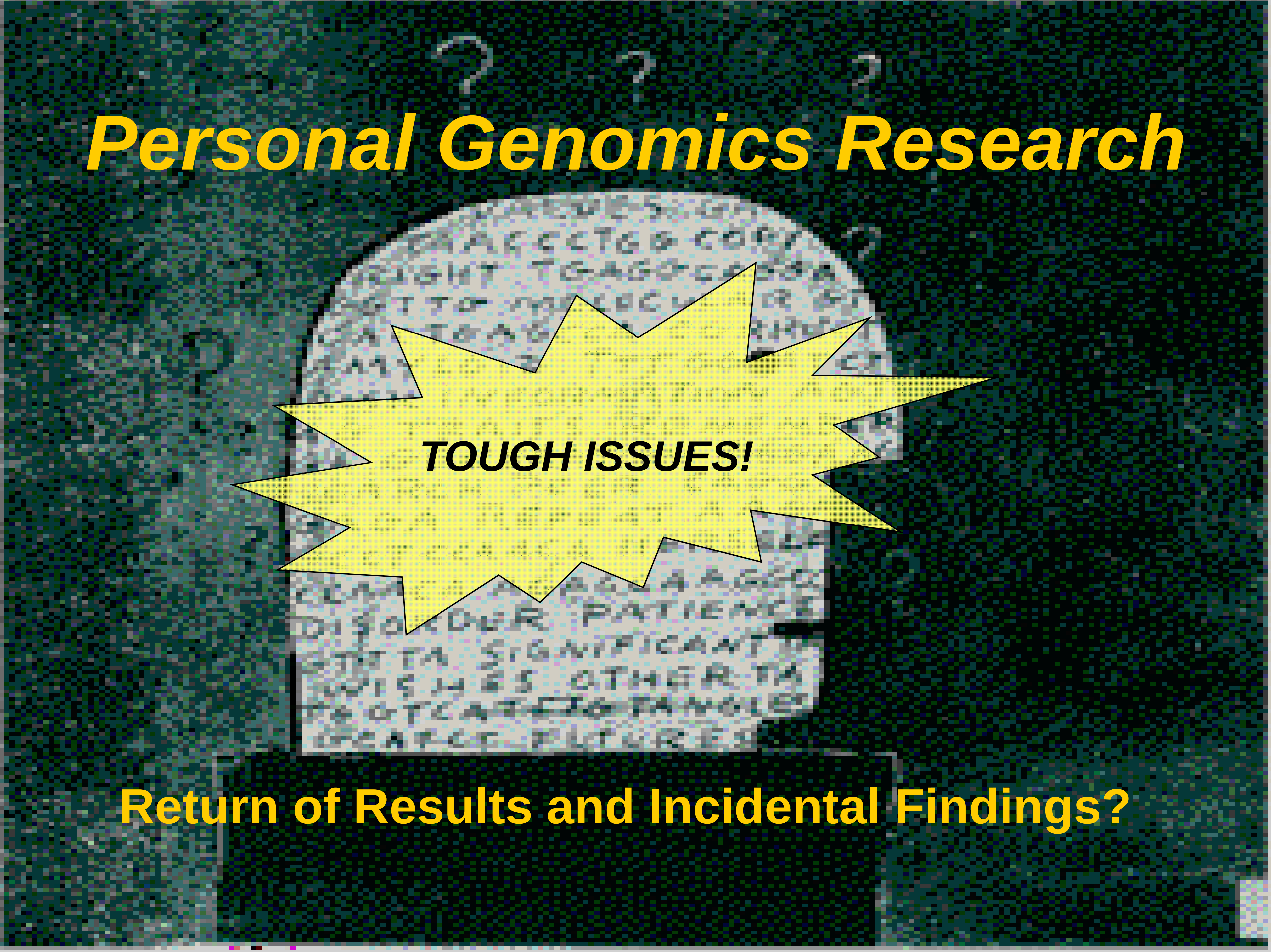
DOI: 10.1080/15265160902928209

Target Article

Social Networkers' Attitudes Toward Direct-to-Consumer Personal Genome Testing

Amy L. McGuire, Center for Medial Ethics and Health Policy, Baylor College of Medicine
Christina M. Diaz, Center for Medial Ethics and Health Policy, Baylor College of Medicine
Tao Wang, Lester & Sue Smith Breast Center, Baylor College of Medicine
Susan G. Hilsenbeck, Lester & Sue Smith Breast Center, Baylor College of Medicine

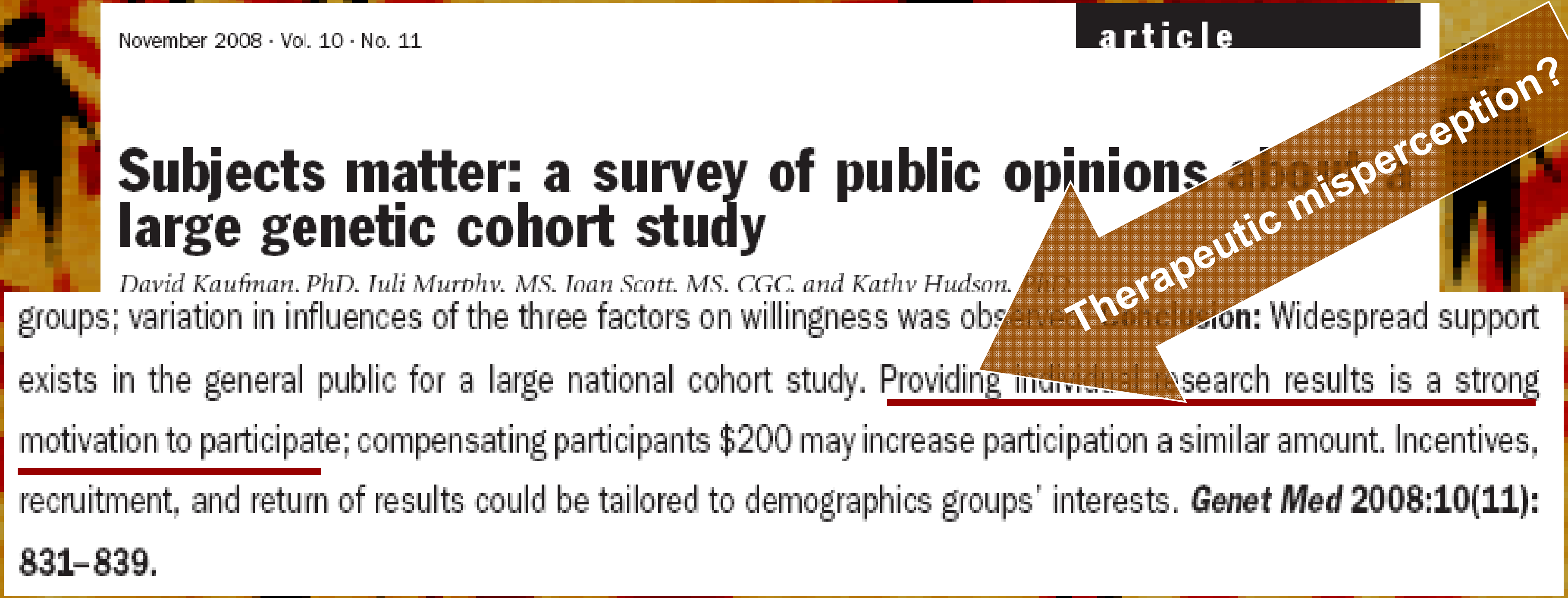
Personal Genomics Research



TOUGH ISSUES!

Return of Results and Incidental Findings?

Do they want results?



November 2008 · Vol. 10 · No. 11

article

Subjects matter: a survey of public opinions about a large genetic cohort study

David Kaufman, PhD, Juli Murphy, MS, Joan Scott, MS, CGC, and Kathy Hudson, PhD

groups; variation in influences of the three factors on willingness was observed. **Conclusion:** Widespread support exists in the general public for a large national cohort study. Providing individual research results is a strong motivation to participate; compensating participants $200 may increase participation a similar amount. Incentives, recruitment, and return of results could be tailored to demographics groups' interests. ***Genet Med*** **2008:10(11): 831–839.**

Therapeutic misperception?

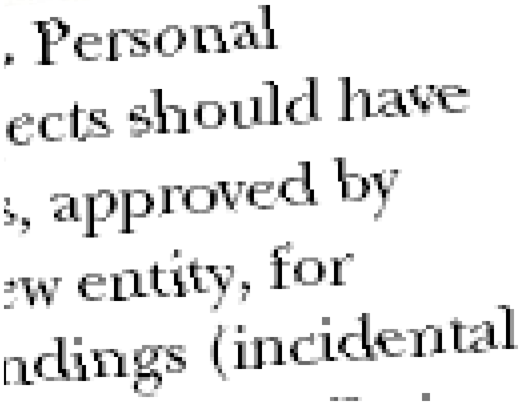
Legal obligation?
Interest in results driven by pop culture representations?
What do you return?

Legal Norms?

The Diploid Genome Sequence of J. Craig V

Managing Incidental Findings in Human Subjects Research: Analysis and Recommendations

Susan M. Wolf, Frances P. Lawrenz, Charles A. Nelson, Jeffrey P. Kahn, Mildred K. Cho, Ellen Wright Clayton, Joel G. Fletcher, Michael K. Georgieff, Dale Hammerschmidt, Kathy Hudson, Judy Illes, Vivek Kapur, Moira A. Keane, Barbara A. Koenig, Bonnie S. LeRoy, Elizabeth G. McFarland, Jordan Paradise, Lisa S. Parker, Sharon F. Terry, Brian Van Ness, and Benjamin S. Wilfond



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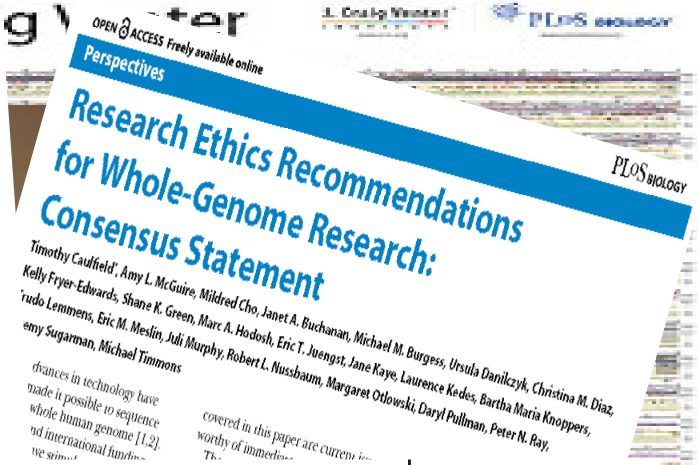


OPEN ACCESS Freely available online

Perspectives

PLoS BIOLOGY

Research Ethics Recommendations for Whole-Genome Research: Consensus Statement

Timothy Caulfield*, Amy L. McGuire, Mildred Cho, Janet A. Buchanan, Michael M. Burgess, Ursula Danilczyk, Christina M. Diaz, Kelly Fryer-Edwards, Shane K. Green, Marc A. Hodosh, Eric T. Juengst, Jane Kaye, Laurence Kedes, Bartha Maria Knoppers, rudo Lemmens, Eric M. Meslin, Juli Murphy, Robert L. Nussbaum, Margaret Otlowski, Daryl Pullman, Peter N. Ray, emy Sugarman, Michael Timmons

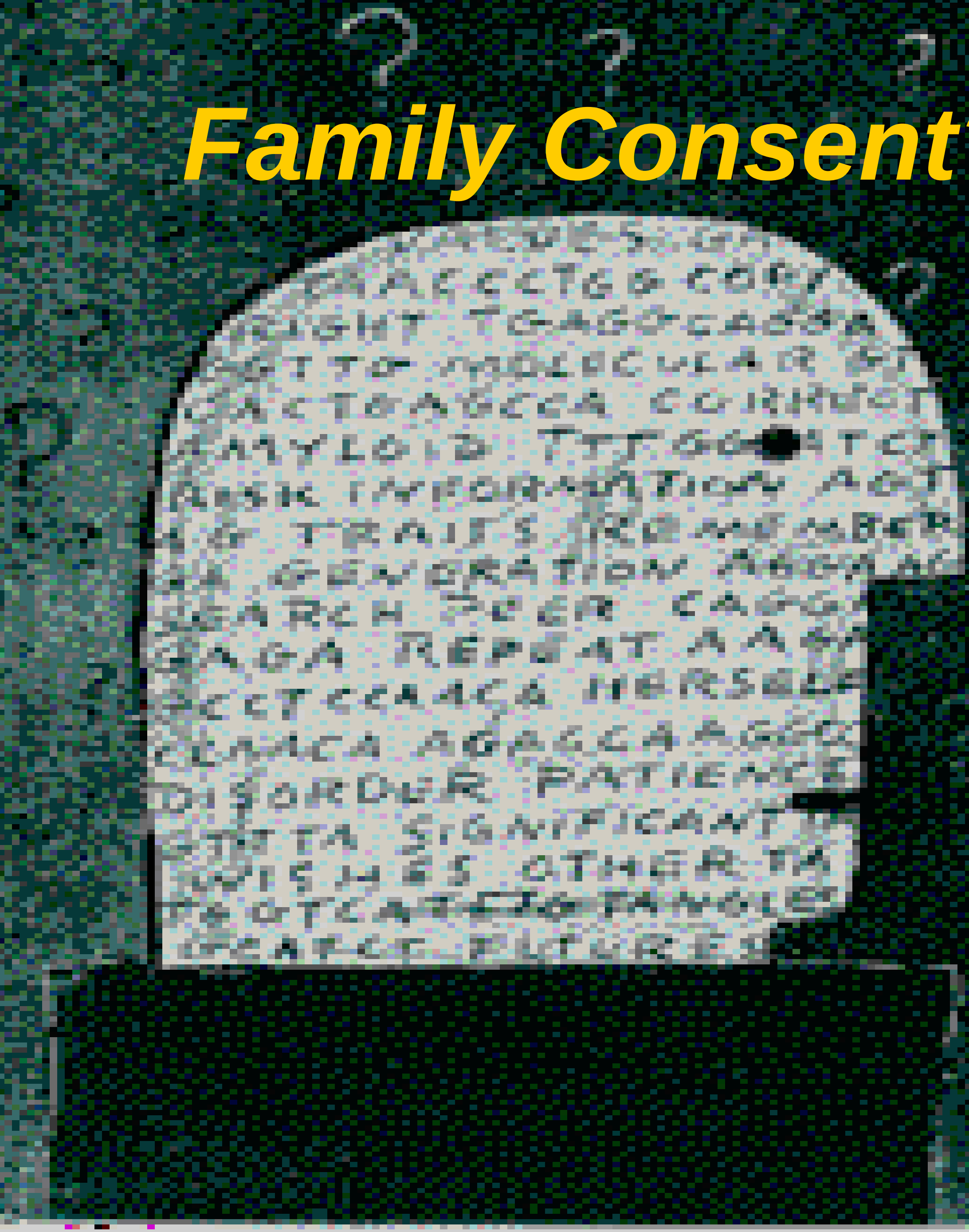
dvances in technology have made it possible to sequence whole human genome [1,2]. nd international funding we stimul

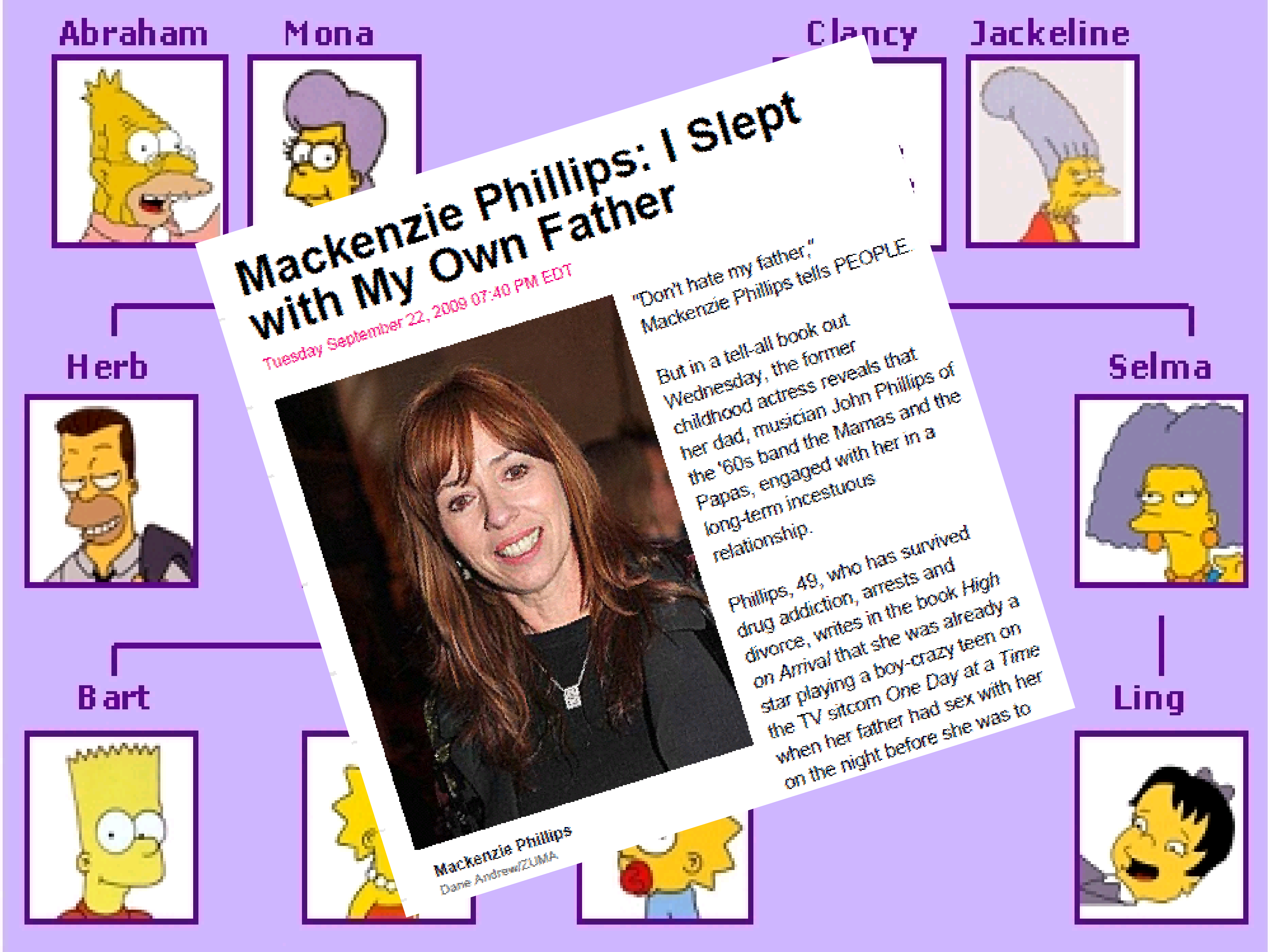
covered in this paper are current is worthy of immedia Th

Our analysis of key ethics sources concludes that researchers have an obligation to address the possibility of discovering IFs not only in their protocol and communications with the IRB, but also in their consent forms and communications with those being recruited to the study and research participants.

mc ... rnal.pbio.0050254.sd001, Levy et al, 2007

Family Consent?





Mackenzie Phillips: I Slept with My Own Father

Tuesday September 22, 2009 07:40 PM EDT

Mackenzie Phillips

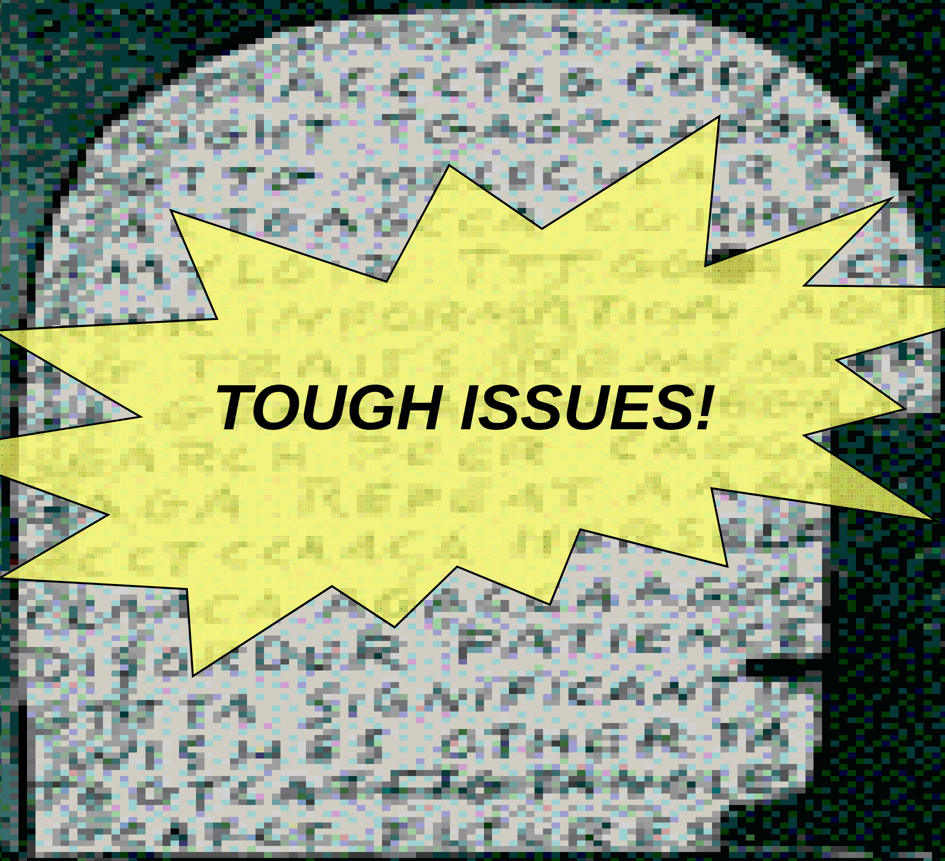
Dane Andrew/ZUMA

"Don't hate my father," Mackenzie Phillips tells PEOPLE.

But in a tell-all book out Wednesday, the former childhood actress reveals that her dad, musician John Phillips of the '60s band the Mamas and the Papas, engaged with her in a long-term incestuous relationship.

Phillips, 49, who has survived drug addiction, arrests and divorce, writes in the book *High on Arrival* that she was already a star playing a boy-crazy teen on the TV sitcom *One Day at a Time* when her father had sex with her on the night before she was to

Patent Problems?



TOUGH ISSUES!

The access and "anti-commons" debate

Judge not thrilled with idea of patenting human genes…

Plaintiffs have cited other studies to demonstrate the chilling effect of gene patents on the advancement of both genetic research and clinical testing. A survey of laboratory directors in the United States conducted by Dr. Mildred Cho (the "Cho study") found that 53% decided not to develop a new clinical test because of a gene patent [illegible] license, [illegible] believed that gene [illegible] the [illegible] research [illegible] ¶ 10. This correlated with a study conducted by the American [illegible] of [illegible] of respondents felt that patents had delayed [illegible] limited their research. Cho Decl. ¶ 11. The Cho study also revealed

Federal Court Rules that Myriad's BRCA Paten [illegible] DNA' Unpatentable

March 30, 2010

http://www.genomeweb.com/dxpgx/federal-court-rules-myriads-brca-patents-are-invalid-deems-isol

NEW YORK (GenomeWeb News) – [illegible] court today sided with the American Civil Li

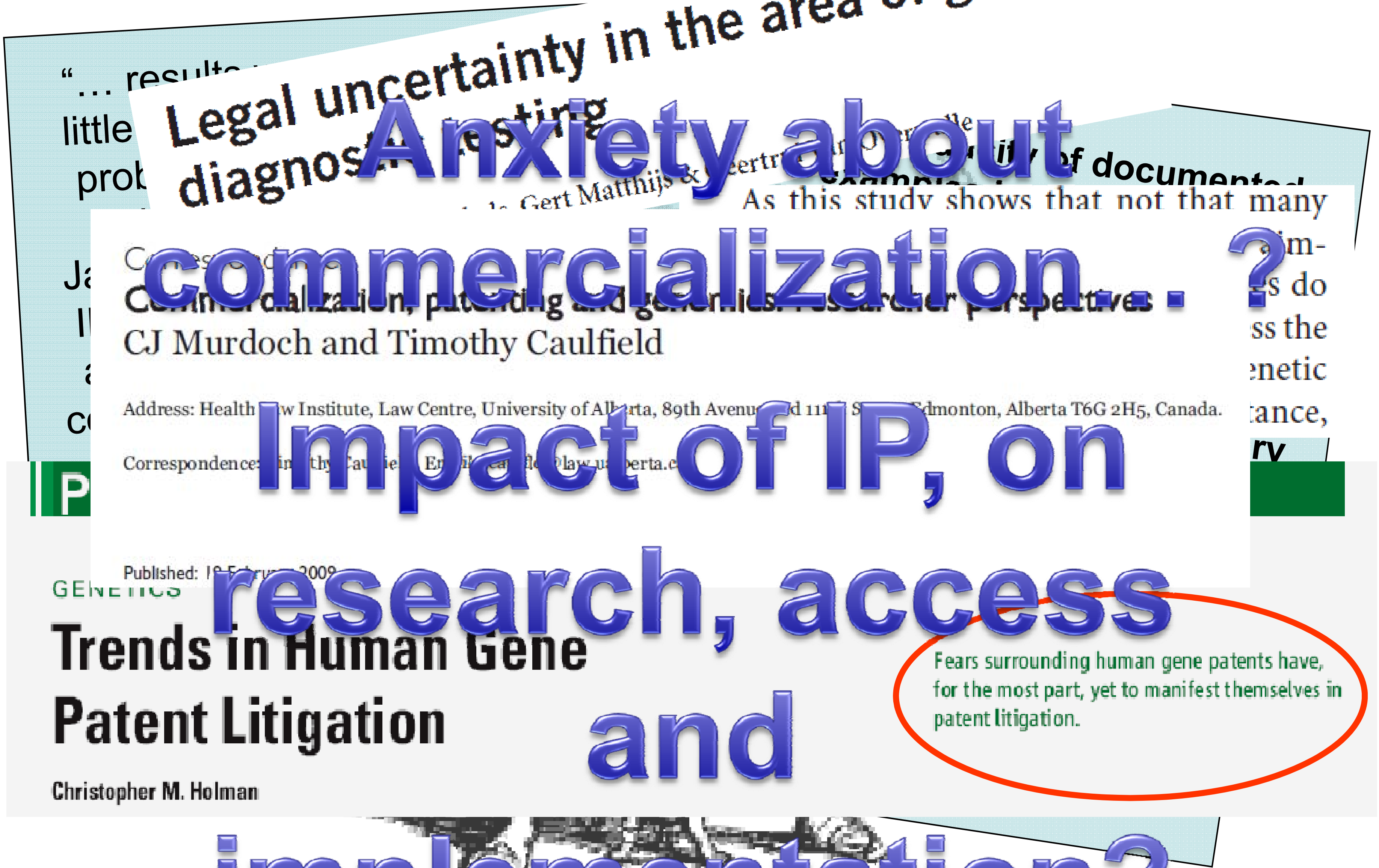
plaintiffs in an ongoing anti-ge [illegible] declaring Myriad Genetics' BRCA

March 30, 2010

After Patent on Genes Is Invalidated, Taking Stock

By ANDREW POLLACK

Little Evidence to Support C…n…



"… results …
little …
prob…

Legal uncertainty in the area of genetic diagnostic testing

Gert Matthijs &

…ity of documented examples

As this study shows that not that many
…im-
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Commercialization, patenting and genomics: researcher perspectives

CJ Murdoch and Timothy Caulfield

Address: Health Law Institute, Law Centre, University of Alberta, 89th Avenue and 111 Street, Edmonton, Alberta T6G 2H5, Canada.

Correspondence: Timothy Caulfield …

Published: 19 February 2009

GENETICS

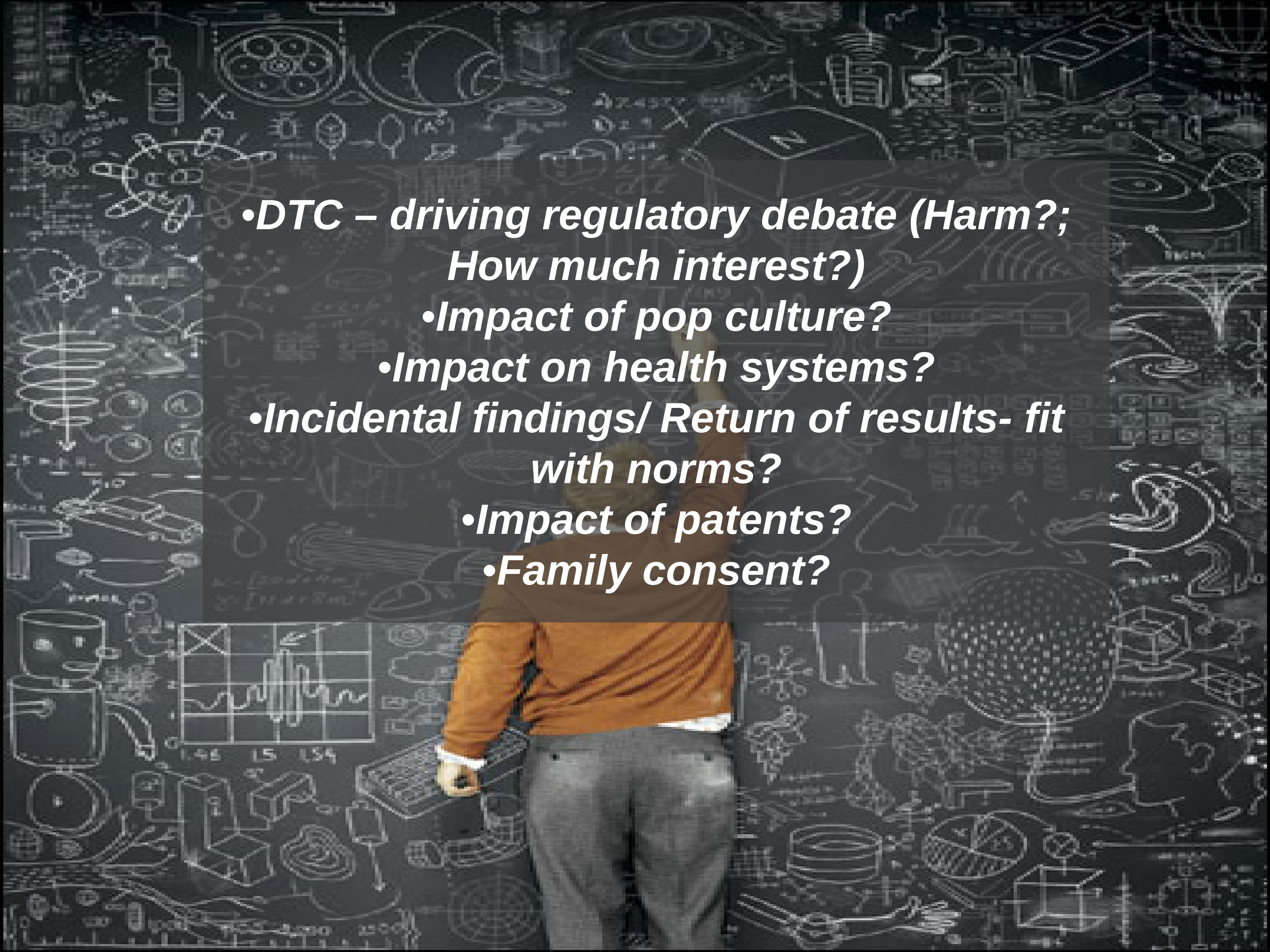
Trends in Human Gene Patent Litigation

Christopher M. Holman

Fears surrounding human gene patents have, for the most part, yet to manifest themselves in patent litigation.

Anxiety about commercialization… ?

Impact of IP, on research, access and implementation?



- ***DTC – driving regulatory debate (Harm?; How much interest?)***
- ***Impact of pop culture?***
- ***Impact on health systems?***
- ***Incidental findings/ Return of results- fit with norms?***
- ***Impact of patents?***
- ***Family consent?***

Thank you!

- Amy Zarzeczny, Robyn Hyde-Lay, Ubaka Ogbogu, Nola Ries, Tania Bubela, the AHFMR, and the HLI research team.

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