

Best Practices in Personalized Medicine
Vancouver, BC
March 8-9, 2011

“Workable” Ethics for Translational Research?

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Research Ethics

TCPS2

TRI-COUNCIL POLICY STATEMENT

Ethical Conduct for Research Involving Humans

2010

Canadian Institutes of Health Research
Natural Sciences and Engineering Research Council of Canada
Social Sciences and Humanities Research Council of Canada

Please cite this document as follows:

Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada, and Social Sciences and Humanities Research Council of Canada, *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans*, December 2010.

Note: For the most recent information on amendments, please consult the official online version of the TCPS at www.pre.ethics.gc.ca.

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Catalogue No. MR23-10/2010E-PDF
ISBN 978-1-100-17237-01

Clinical Trials

Guidance for Industry

Clinical Pharmacogenomics: Premarketing Evaluation in Early Phase Clinical Studies

DRAFT GUIDANCE

This guidance document is being distributed for comment purposes only.

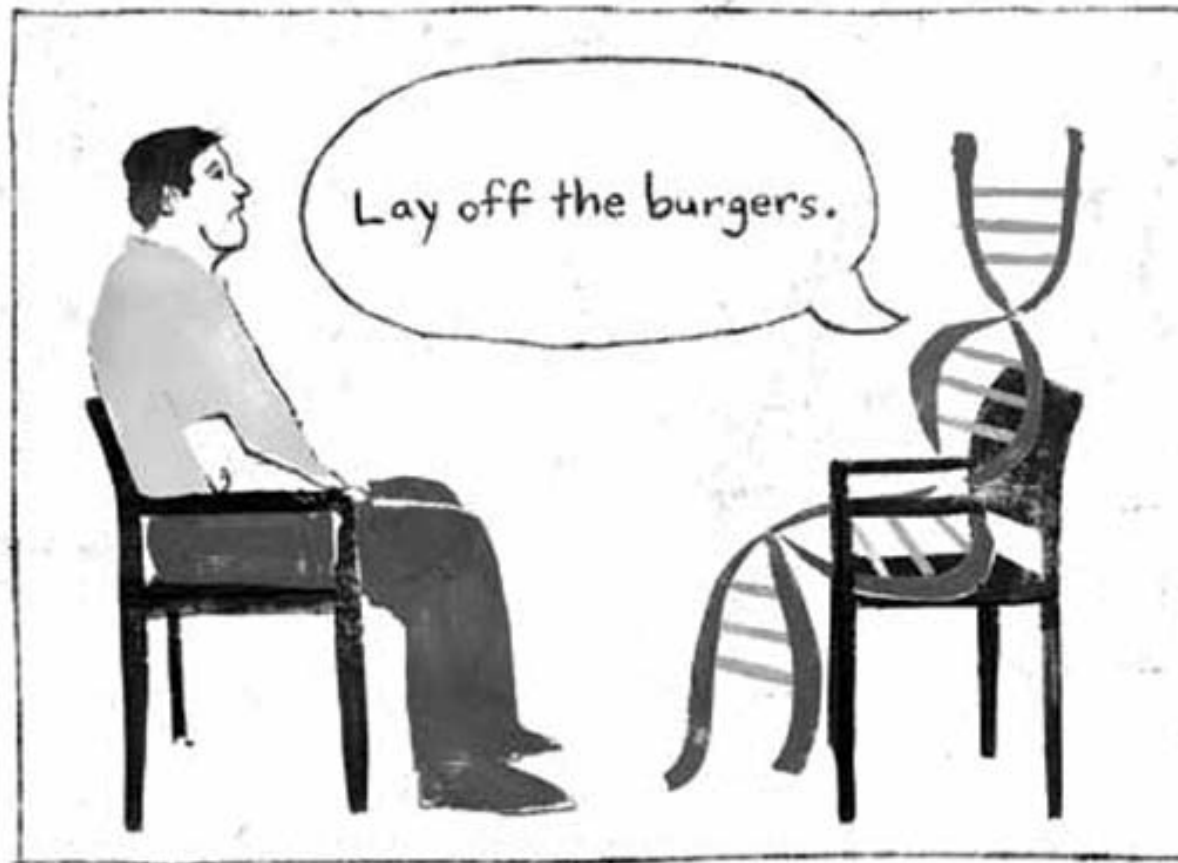
Comments and suggestions regarding this draft document should be submitted within 60 days of publication in the *Federal Register* of the notice announcing the availability of the draft guidance. Submit comments to the Division of Dockets Management (HFA-301), Food and Drug Administration, 5630 Fishers Lane, rm. 1061, Rockville, MD 20852. All comments should be identified with the docket number listed in the notice of availability that publishes in the *Federal Register*.

For questions regarding this draft document, contact (CDER) Lawrence Lesko at 301-796-1565 or Shiew-Mei Huang at 301-796-1541, or (CBER) Office of Communication, Outreach and Development at 800-835-4709 or 301-827-1800, or Changting Haudenschild at 301-827-3947, or (CDRH) Frances Kalush at 301-796-5408.

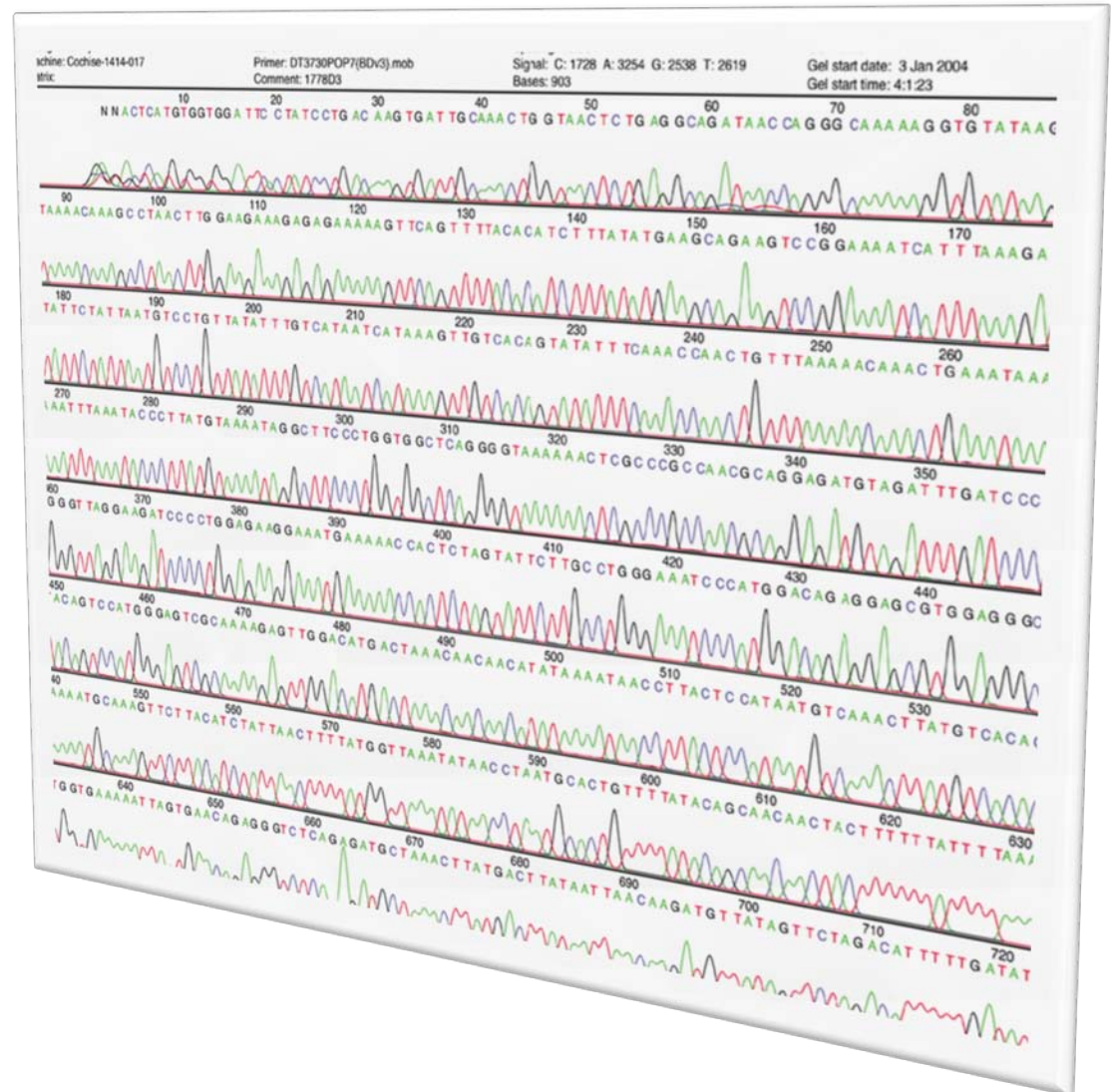
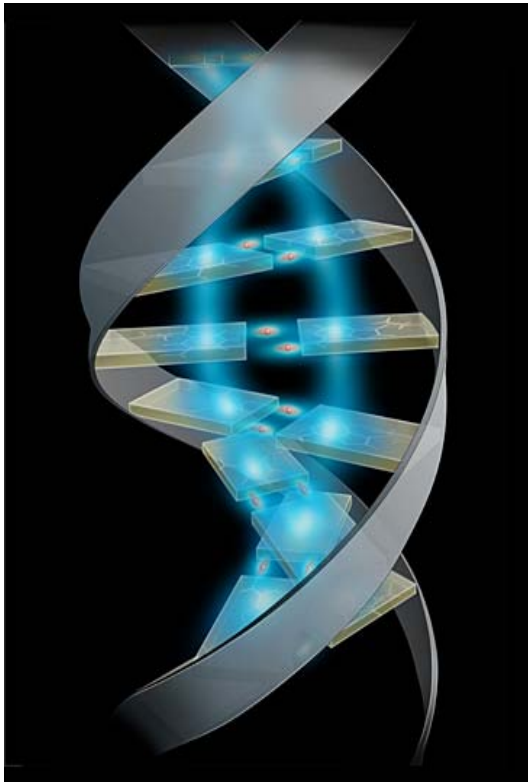
U.S. Department of Health and Human Services
Food and Drug Administration
Center for Drug Evaluation and Research (CDER)
Center for Biologics Evaluation and Research (CBER)
Center for Devices and Radiological Health (CDRH)

February 2011
Clinical Pharmacology

Physician – Patient Relationship



Whole-Genome Sequencing



Population Biobanks

POLICYFORUM

RESEARCH FUNDING

Sustaining the Data and Bioresource Commons

Paul N. Schofield,^{1,2*} Janan Eppig,² Eva Huala,³ Martin Hrabe de Angelis,⁴ Mark Harvey,⁵ Duncan Davidson,⁶ Tom Weaver,⁷ Steve Brown,⁸ Damian Smedley,⁹ Nadia Rosenthal,¹⁰ Klaus Schughart,¹¹ Vassilis Aidinis,¹² Glauco Tocchini-Valentini,¹³ John M. Hancock⁸

Globalization of biomedical research requires sustained investment for databases and biorepositories.

Development of powerful, high-throughput technologies, together with globalization of scientific research, presents the biomedical research community with unprecedented challenges for the management, archiving, and distribution of data and bioresources (*1*). We need a social contract between funding agencies and the scientific community to accommodate “bottom-up” integration and “top-down” financing of databases and biorepositories on an international scale.

The biological commons is evolving away from a traditional differen-



data models to accommodate new data types. The associated bioontologies and other informatics tools need to continue to be developed, maintained, and applied to data to standardize and maximize access, retrieval, and exploitation for discovery. Repositories also need to innovate and respond to emerging disruptive technologies. Consequently, any distinction between time-delimited research projects and long-term, relatively static infrastructures is being eroded. The traditional distinction between “infrastructure” and “research” is even less appropriate, presenting a challenge to those funders who continue to think in these

II. IMPLICATIONS

A. Recruitment

HapMap and 1000Genomes

International HapMap Project

Home | About the Project | Data | Publications | Tutorial

English | Français | Yoruba

The International HapMap Project is a partnership of scientists and funding agencies from Canada, China, Japan, Nigeria, the United Kingdom and the United States to develop a public resource that will help researchers find genes associated with human disease and response to pharmaceuticals. See "About the International HapMap Project" for more information.

Project Information

- About the Project
- HapMap Publications
- HapMap Tutorial
- HapMap Mailing List

News

- 2011-02-02: **Haploview issues with rel 28 data**

Recently, there are several questions about Haploview data format errors when users tried to analyze HapMap release 28 data. The current Haploview version (4.2) does not recognize the new individuals in release 28 and the software will generate an error similar to "Hapmap data format error: NA18876" when trying to open the data.

1000 Genomes

A Deep Catalog of Human Genetic Variation

Home | About | Data | Analysis | Participants | Contact | Browser | Wiki

Search

LATEST ANNOUNCEMENTS

WEDNESDAY FEBRUARY 16, 2011

February 2011 Data Update

Full Project Indel Release

Indels calls from Dindel. These calls are based on 629 individuals from the 20100804 sequence and alignment release of the 1000 genomes project. This release is based on the GRCh37 assembly of the human genome and are released in the format VCF 4.0

Data access links: [EBI/NCBI](#)

Link to additional information: [README file](#)

THURSDAY FEBRUARY 23, 2011

Mapping copy number variation by population scale genome sequencing

The Structural Variant group from the 1000 Genomes Project Consortium has published its findings based on the pilot data analysis in Nature today. [Mapping copy number variation by population scale genome sequencing](#). The data supporting this paper can be found on the [ftp site EBI/NCBI](#)

Recent project announcements

WEDNESDAY FEBRUARY 16, 2011

Release of Full Project Phase 1 Alignments

The alignments based on the 20101123.sequence.index have been released. There are both new BAM files and updated BAM files with more data were added. For the case of updated files, the older, redundant files have been withdrawn.

Illumina data was aligned at the Sanger Institute and SOLiD data at TGEN. Full project data is aligned to the GRCh37 human assembly.

Data access links: [EBI/NCBI](#) / [Instructions for data download and Aspera](#)

LINKS

- All Project Announcements
- Sample and Project Information
- Media Archive
- Download the 1000 Genomes Pilot Paper
- Project Contacts
- RSS Feed

A. Recruitment

"Citizens" and Population Biobanks

TAKING IT TO THE **BioBANK**
By CATHERINE HARRIS



II. IMPLICATIONS

A. Recruitment

Genotyping and Clinical Trials

Commentary

Ethical challenges in genotype-driven research recruitment

Laura M. Beskow,^{1,4} Kristen N. Linney,² Rodney A. Radtke,³ Erin L. Heinzen,² and David B. Goldstein²

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Many genetic and genomic studies are conducted using a phenotype-driven approach: Cases and controls are identified based on the presence or absence of a particular condition and analyses are undertaken to identify gene variants associated with that condition. The inverse—a genotype-driven approach—is receiving increasing attention as another powerful research tool. In this

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II. IMPLICATIONS

A. Recruitment

Personal Genome Project

Personal Genome Project

Home [Project Overview](#) [Participation Overview](#) [PGP Community](#) [DONATE](#)

Volunteers from the general public working together with researchers to advance personal genomics.

We believe individuals from the general public have a vital role to play in making personal genomes useful. We are recruiting volunteers who are willing to share their genome sequence and many types of personal information with the research community and the general public, so that together we will be better able to advance our understanding of genetic and environmental contributions to human traits and to improve our ability to diagnose, treat, and prevent illness. Learn more about how to [participate](#) in the Personal Genome Project.



Project Overview. The PGP hopes to make personal genome sequencing more affordable, accessible, and useful for humankind. Learn more about our [mission](#).



Want to participate? We aim to enroll 100,000 informed participants from the general public. Learn more about [participation](#) in the PGP and how you can get involved.



Meet our volunteers. Participants may volunteer to publicly share their DNA sequence and other personal information for research and education. Meet the "[PGP-1K](#)".



Documentary Film about PGP. Two-time Emmy Award-winning documentary producer [Marilyn Ness](#) is making a film about the PGP.



CC0. We are committed to making [research data](#) from the PGP freely available to the public. Read about PersonalGenomes.org's use of the



Help Us Build. Are you a Rails developer interested in supporting the PGP through volunteer web application development? See our

Project News

Keep up-to-date with PersonalGenomes.org news and events. [Subscribe to our newsletter](#).

November 9, 2010: George Church wins Franklin Institute Bower Prize. [Read news](#).

October 12, 2010: Announcing new collaborations, new tools, and the PGP-1K. [Read newsletter #4](#).

October 11, 2010: Welcome to the team, [PGP-1K](#)! This week we are enrolling the next 1000 participants in the PGP.

July 2, 2010: New eligibility screening and enrollment software launched. We removed the need for invitation codes. [Sign up now](#).

April 27, 2010: A majority of individuals with a personal genome sequence on one stage at the [GET Conference 2010](#).

II. IMPLICATIONS

B. Privacy

Controlled Access Databases (ICGC)



**International
Cancer Genome
Consortium**

Int. J. Epidemiol. Advance Access published July 14, 2010

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International Journal of Epidemiology 2010;1–11
doi:10.1093/ije/dyq111

DataSHIELD: resolving a conflict in contemporary bioscience—performing a pooled analysis of individual-level data without sharing the data

Michael Wolfson,¹ Susan E Wallace,^{2,3} Nicholas Masca,⁴ Geoff Rowe,¹ Nuala A Sheehan,⁴ Vincent Ferretti,^{3,5} Philippe LaFlamme,^{3,6} Martin D Tobin,⁴ John Macleod,⁷ Julian Little,^{3,8} Isabel Fortier,^{3,8,9} Bartha M Knoppers^{2,3} and Paul R Burton^{3,4,8,10*}

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Accepted 27 May 2010

Background Contemporary bioscience sometimes demands vast sample sizes and there is often then no choice but to synthesize data across several studies and to undertake an appropriate pooled analysis. This same need is also faced in health-services and socio-economic research. When a pooled analysis is required, analytic efficiency and flexibility are often best served by combining the individual-level data

Downloaded from ije.oxfordjournals.org at McGill University Libraries on July 14, 2010

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Downloaded from ije.oxfordjournals.org at McGill University Libraries on

Identification?

OPEN ACCESS Freely available online

PLoS GENETICS

Viewpoints

Public Access to Genome-Wide Data: Five Views on Balancing Research with Privacy and Protection

P³G Consortium^{1*}, George Church^{1*}, Catherine Heeney^{2*}, Naomi Hawkins², Jantina de Vries², Paula Boddington², Jane Kaye², Martin Bobrow^{3*}, Bruce Weir^{4*}

1 Department of Genetics, Harvard Medical School, Cambridge, Massachusetts, United States of America, **2** The Ethox Centre, Department of Public Health and Primary Care, University of Oxford, Oxford, United Kingdom, **3** Department of Medical Genetics, University of Cambridge, Cambridge, United Kingdom, **4** Department of Biostatistics, University of Washington, Seattle, Washington, United States of America

Introduction by Greg Gibson and Elizabeth Fisher

Just over twelve months ago, *PLoS Genetics* published a paper [1] demonstrating that, given genome-wide genotype data from an individual, it is, in principle, possible to ascertain whether that individual is a member of a larger group defined solely by aggregate genotype frequencies, such as a forensic sample or a cohort of participants in a genome-wide association study (GWAS). As a consequence, the National Institutes of Health (NIH) and Wellcome Trust agreed to shut down public access not just to individual geno-

research. George Church posits that actions directed toward restricting data access are likely to exclude researchers who might provide the most novel insights into the data and instead makes the argument that full disclosure and consent to the release of genomic information should be sought from study participants, rather than making difficult-to-guarantee promises of anonymity. Martin Bobrow weighs the risks and benefits and proposes four steps that represent a middle ground: Retain restricted access for now, make malicious de-identification practices illegal, increase public awareness of the issues, and encourage recognition that scientists

DNA mixture including up to 1,000 participants. Although in hindsight it is clear that basic statistical theory would predict this to be the case, the reality is that it had previously gone completely unrecognised.

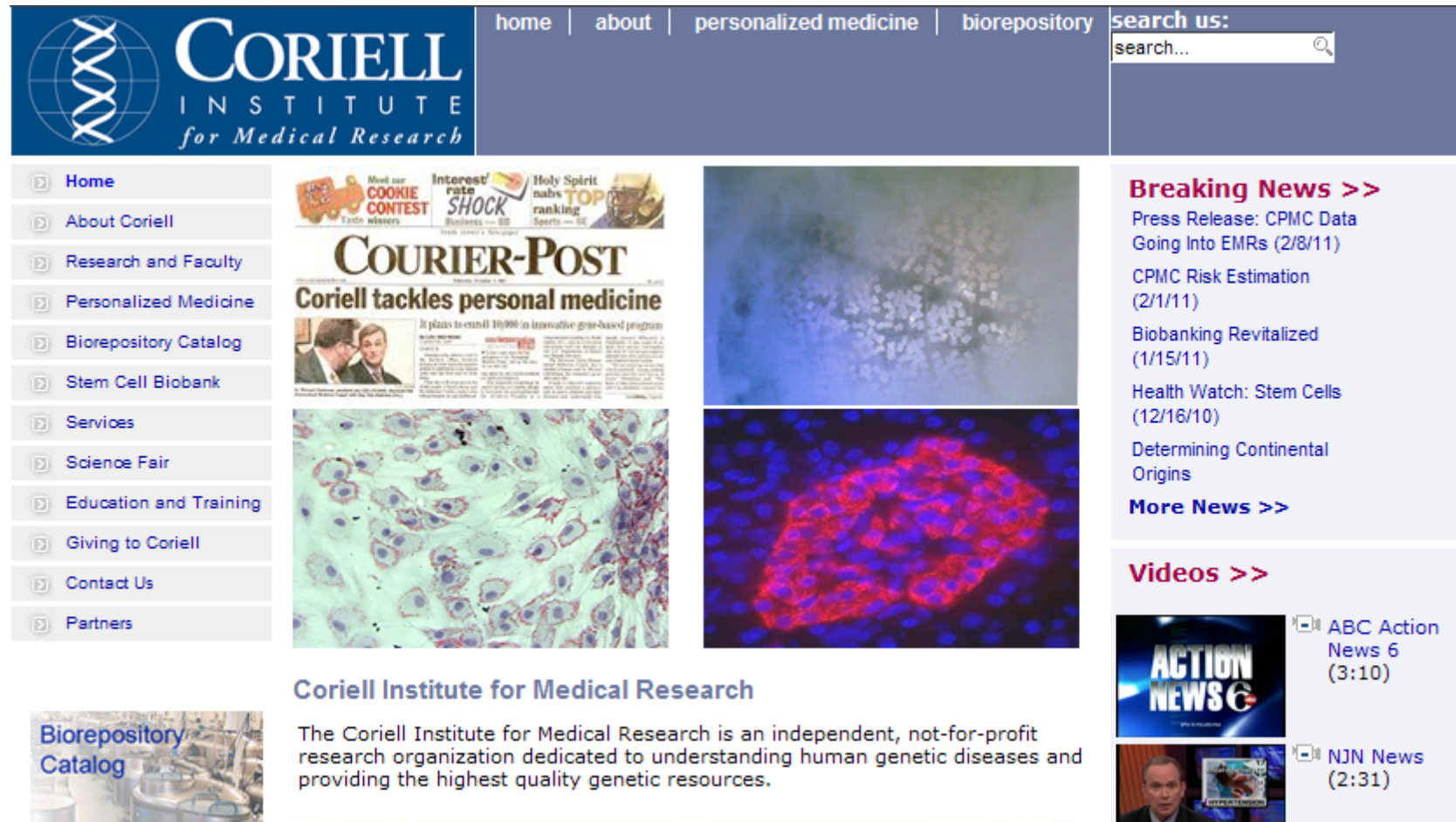
This situation illustrates the need to raise the level of discussion, thereby avoiding the ad hoc resolution of immediate privacy concerns and anticipating future scientific possibilities with a view to providing prospective guidance.

The implications of the Homer paper were discussed by the international Public Population Project in Genomics (P³G) (<http://www.p3g.org>). The consensus was (publicly available) [2]. The consensus was a robustly agreed set of principles (P³G) that addressed the key issues raised by the Homer paper. The substance of the Homer paper is to biomarker biobanking literature. Public scientific biobanking is a key UK biotech corridor and public biobanking is a key part of the UK's research infrastructure. The paper highlights the need to

II. IMPLICATIONS

C. Communication

Biobanks Websites



CORIELL
INSTITUTE
for Medical Research

home | about | personalized medicine | biorepository

search us:
search...

Home
About Coriell
Research and Faculty
Personalized Medicine
Biorepository Catalog
Stem Cell Biobank
Services
Science Fair
Education and Training
Giving to Coriell
Contact Us
Partners

COURIER-POST
Coriell tackles personal medicine
It plans to enroll 10,000 in innovative gene-based program

Breaking News >>
Press Release: CPMC Data Going Into EMRs (2/8/11)
CPMC Risk Estimation (2/1/11)
Biobanking Revitalized (1/15/11)
Health Watch: Stem Cells (12/16/10)
Determining Continental Origins
More News >>

Videos >>
ABC Action News 6 (3:10)
NJN News (2:31)

Biorepository Catalog

Coriell Institute for Medical Research
The Coriell Institute for Medical Research is an independent, not-for-profit research organization dedicated to understanding human genetic diseases and providing the highest quality genetic resources.

II. IMPLICATIONS

C. Communication

Return of Results?



McGill



CENTRE OF GENOMICS AND POLICY
CENTRE DE GÉNOMIQUE ET POLITIQUES

THE JOURNAL OF
LAW, MEDICINE & ETHICS

A Journal of the American Society of Law, Medicine & Ethics

Lexicon

International Context and General Principles

Biobanks and Populations

Paediatric Research

Intrafamilial Communication

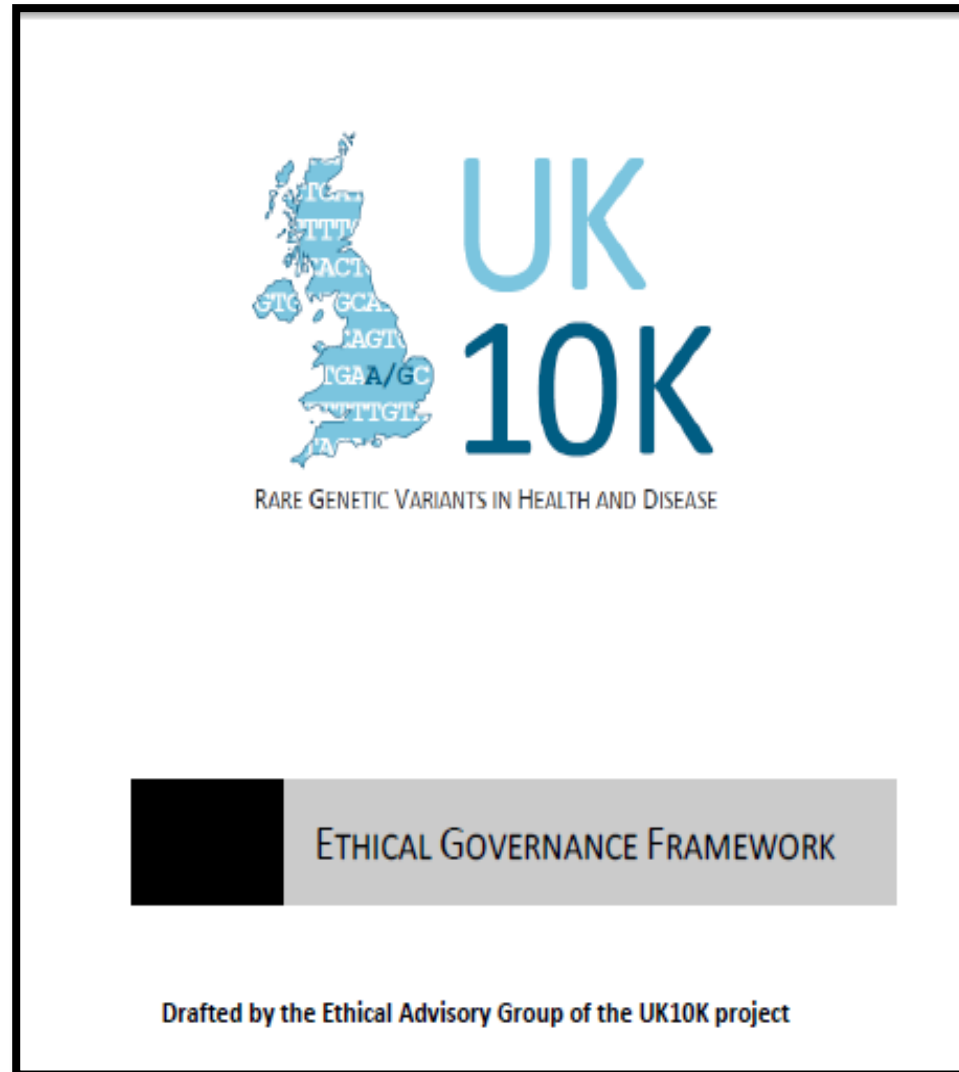
Deceased Individuals

International Consortia

II. IMPLICATIONS

C. Communication

“Incidental” Findings



Data-Sharing

International Code of Conduct for Data Sharing in
Genomic Research



Genomic Research: Quo Vadis?

PERSPECTIVE

doi:10.1038/nature09764

Charting a course for genomic medicine from base pairs to bedside

Eric D. Green¹, Mark S. Guyer¹ & National Human Genome Research Institute*

There has been much progress in genomics in the ten years since a draft sequence of the human genome was published. Opportunities for understanding health and disease are now unprecedented, as advances in genomics are harnessed to obtain robust foundational knowledge about the structure and function of the human genome and about the genetic contributions to human health and disease. Here we articulate a 2011 vision for the future of genomics research and describe the path towards an era of genomic medicine.

CONCLUSION

