

Australasian Science

Volume 37 | Number 6
JULY/AUGUST 2016 | \$9.95

SPECIAL ISSUE

Genomics

**Do Genome Tests
Live up to the Hype?**

**Are We Prepared for
Our Test Results?**

**Should Genomes Be
Screened at Birth?**

**Should You Share Your
Genetic Data Online?**

**Will a Genome Test Lead
to Healthy Behaviours?**

**Indigenous Questions
about Genomic Research**

**Global Biobanks: A New
Frontier for Big Data?**

**The Changing Landscape
of Gene Patent Law**

The Stem Cell "Sell"

Toxic Legacy of Firefighting Foam • Mindless Scientology • Linguistic Diagnosis of Disease

ISSN 1442-679X



9 771442 679017

FEATURES

- 12 Genomic Testing as a Lifetime Health Resource?**
If lives could be saved by being “forewarned” by a genomic test, should we perform genomic testing of all babies at birth?
- 15 Will Genomics Motivate Healthy Behaviours?**
Will communicating the genetic risks of disease necessarily motivate people to make healthier behaviour choices?
- 18 Indigenous Genomics**
Mistrust is a significant but not insurmountable barrier to the acceptance of genomics by Indigenous people.
- 21 The Ethics of Online Genomics Tests**
There is a significant difference between the expectation and reality of direct-to-consumer personal genome testing, creating a gap where interesting tensions and ethical dilemmas sit.
- 24 Like, Comment, Share: Should You Share Your Genetic Data Online?**
The culture of sharing our private details online is extending to health and ancestry data generated by genome testing. What are the benefits and what are the risks?
- 27 Personal Genomics: What Do Consumers Want?**
Are Australian consumers excited or cynical about the promises of personal genome tests, and are they adequately prepared for the information they’ll receive?
- 30 The Changing Role of IP in Genomics**
Recent court decisions have overturned previous rulings about genetic patents, but other intellectual property regimes are already taking their place.
- 33 Biobanks Go Global**
Global networks of depositories for biological samples open a range of scientific, legal and ethical challenges.
- 36 The Stem Cell “Sell”**
The unfettered commercial environment that has allowed stem cell tourism to flourish must be challenged, and the professionals who enable it should be held to account.



conSCIENCE

- 39 A Toxic Legacy from Firefighting Foams**
Australian communities and environmental systems adjacent to Defence sites, airports and firefighting training centres have been contaminated by toxic chemicals.



9

NEWS

6 Browse

A round-up of science news from our shores.

COLUMNS

5 Guest Editorial

This edition of *Australasian Science* focuses on the ethical, legal and social issues associated with advances in genomic science.

40 Expert Opinion

Two reports have reviewed the evidence for the safety of nanotechnologies in food packaging and food additives.

41 Neuropsy

Linguistic analysis can distinguish between genuine and falsified expressions of remorse.

42 The Fit

What if doctors could diagnose disease by what you wrote on Facebook and Twitter?

43 The Fossil File

The public are welcome to attend one of Australia's largest palaeontology conferences.

44 Directions

The mass bleaching on the Great Barrier Reef this summer is the most damaging ever recorded.

45 Out of this World

Astronomers have discovered the closest habitable planet outside our solar system, and discovered one of the biggest black holes ever.

46 The Bitter Pill

Populist TV, blogs and publications have portrayed saturated fats as healthy rather than dietary villains, but this is an oversimplification.

47 The Naked Skeptic

Scientologists argue against the existence of the mind, and therefore the existence of mental illness.

48 EcoLogic

Can automated algorithms do better than humans in conservation games?

49 Lowe Tech

The election revealed a bipartisan lack of understanding of the role of science in innovation and of the coal industry in the fate of the Great Barrier Reef.

50 Quandary

If neural lacing enables our brains to be networked, we could easily be hacked or become the tools of Google or government.

51 Australasian Sky

Your map of the night sky this month.



41



45



50

Please cite as:

Paul H. Mason, Wendy Lipworth & Ian Kerridge, *Biobanks Go Global*, *Australasian Science* July/Aug 2016, available at <http://www.australasianscience.com.au/article/issue-julyaugust-2016/biobanks-go-global.html>



Biobanks Go Global

PAUL H. MASON, WENDY LIPWORTH & IAN KERRIDGE

Global networks of depositories for biological samples open a range of scientific, legal and ethical challenges.

Medical research increasingly relies on collections of donated human tissue, such as DNA samples, blood samples and solid organs and tissues. These collections of donated samples – referred to as biobanks, biorepositories or tissue banks – can be used in basic science experiments, population studies, or towards the refinement and personalisation of medical and surgical techniques.

The practice of collecting and systematically organising biological samples is not new. Famous historical collections were put together by Carl Linnaeus (1707–78) in Sweden, Georges-Louis Leclerc, Comte de Buffon (1707–88) in France, and Joseph Banks (1743–1820) in England.

In recent years, however, advances in experimental techniques (such as whole genome sequencing) and information technologies (such as “big data” storage and analytics) have massively increased the promise of biomedical research using collections of human biological samples.

In order for the promise of biobanks to be fulfilled, large numbers of samples need to be collected, stored and analysed. Until recently, most biobanks were located within individual universities, research institutes or health facilities, and often tied to specific research projects.

Increasingly, however, biobanks have become “networked” in an effort to become more sustainable and to increase their utility. Most often, these networks of biobanks draw together samples from within a country to create a single biobank that

Once a biobank shares samples or biodata with another institution, how can they control or oversee the uses, or on-selling, of that tissue?

may be accessible to researchers based in different institutions and with different research interests.

But even large domestic biobanks or biobank networks like these may still lack the statistical power to answer important research questions. For example, research into the genetics of rare or complex diseases often requires the analysis of samples that number in the hundreds of thousands, if not millions. Maximising the full potential of biobanks is therefore difficult within the confines of a single nation.

As a result, biobanks are now “going global”. This can happen in a number of ways. One possible model is that a single biobank might collect biological samples from numerous countries. Several biobanks in the United States, for instance, use a “direct-to-consumer” model in which they sell genetic testing to customers in numerous countries, and in the process build their biorepositories.

An alternative model is to form virtual networks across national borders through the use of data-sharing technologies and the latest bioinformatic platforms. For example, biobanks in the European Union and the Community of Latin Amer-

ican and Caribbean States have established extensive data-sharing networks that have proven to be useful biodata repositories for tissue samples that are spread across multiple countries.

A third model is for a biobank to distribute its operations, in a compartmentalised way, across numerous countries. For example, a biobank might collect data samples in one country, store these samples in another country and conduct data analysis in yet another location.

While there is enormous excitement about these large biobank networks, the reality is that “going global” is not straightforward. Broadly speaking, barriers to the globalisation of biobanking can be divided into technical issues, ethical issues and economic issues.

Technical issues relate to the sheer size and scale of the bionetworks that are being created, and include challenges associated with collecting, storing, linking and analysing large amounts of data. Participants may need to be recruited at numerous locations to donate samples, undergo a baseline survey and then be followed-up at a later date.

Biobank networks may struggle to ensure that standard scientific practices have been used in every location. Scientific practices, though ostensibly objective and standardised, do not actually exhibit the universal consistency that one might imagine. Whether it's the concentration of reagents, the temperature of storage or the basic equipment used in the laboratory, the technical side of collecting samples for storage can be highly variable.

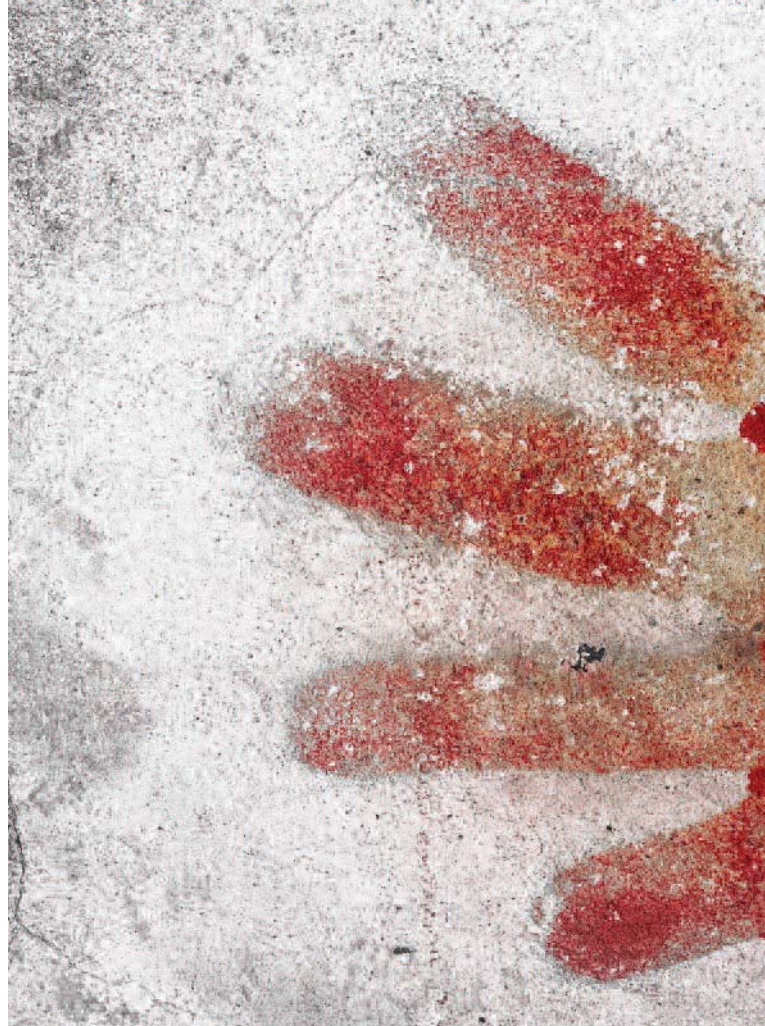
From an ethical perspective, those who want to establish global biobanks have to carefully navigate a whole range of issues including informed consent, confidentiality, control of data access, right to withdraw, accessibility to biospecimens, ownership, benefit sharing, commercialisation, intellectual property rights, genetic discrimination, community engagement, respect for tissue, cultural diversity, beneficence, reciprocity and equity.

Consent refers to informing tissue donors about the research, its associated risks and benefits, and obtaining permission from them to use their tissue. Consent can be narrow for specific research projects or broad, covering a wide range of present and future projects.

Confidentiality refers to the need to protect the anonymity of the donors who have donated samples. Information associated with or derived from a sample may reveal deep insights into individuals' personal lives, and thus privacy is a key issue.

Ownership of tissue also needs to be negotiated carefully, not just between donors and the biobank but also between biobanks and the researchers studying and analysing the samples.

A concern for benefit sharing means that the distribution of benefits from the findings of biobank research are equitably shared within and across populations, with due consideration to the donors who provided the samples.

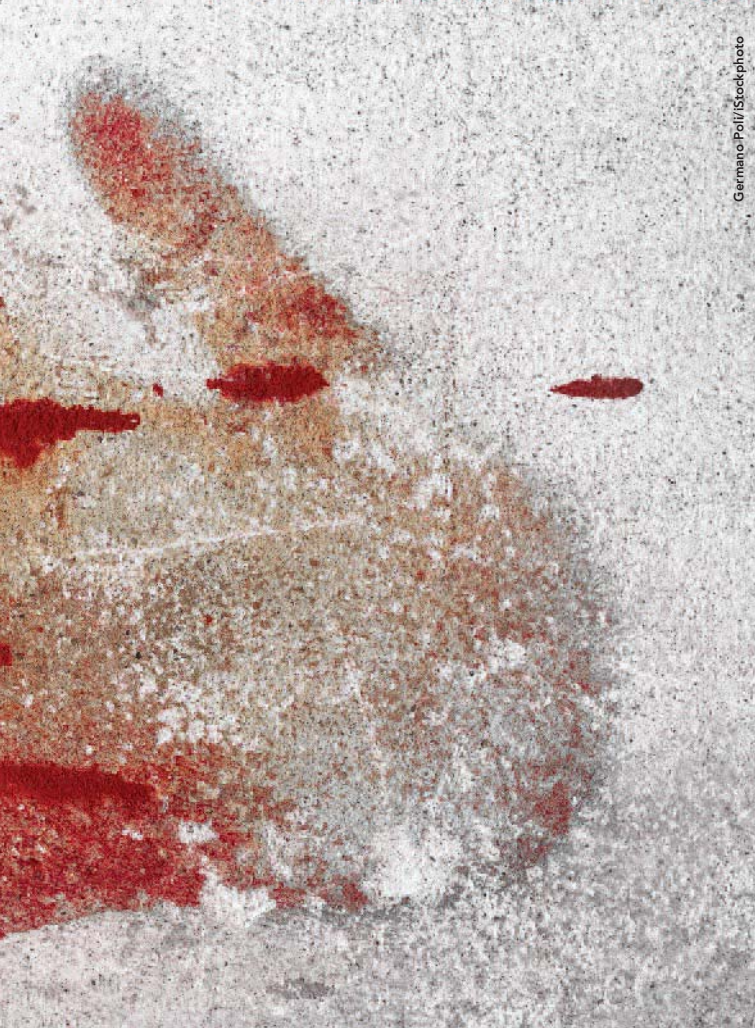


Respect for the samples means using and disposing of tissues in accordance with the values, beliefs and preferences of the donor. Observing these ethical principles locally is an important part of establishing and maintaining trust between a biobank and the general public.

“Going global” not only magnifies existing ethical issues associated with collecting human biological samples, but also opens up a range of new ethical considerations. A key issue that arises as biobanks globalise is the custodianship of tissue. Once a biobank shares samples or biodata with another institution, how can they control or oversee the uses, or on-selling, of that tissue? This matters because the networking of samples opens the space for uses by unknown and possibly unregulated groups whose projects may not be covered by the original research ethics approval and donor consent. The transfer of samples across national boundaries also creates challenges to do with access to samples.

The right to withdraw from research participation without any consequences is standard practice in contemporary biomedical research. This raises the question of what, if any, recourse do donors have to ensure the destruction of their sample if it has been transported abroad?

Even more ethical issues are raised when inequalities between low-income and high-income countries are considered. Countries already dominant in the field are currently setting the research agenda and financial feasibility of global biobanks. If donor tissue is exported from the developing world to the developed world, could people in low-income populations be



Germano Polli/istockphoto

The competing demands of donors, investors, researchers, medical practitioners, patients, families and others who might benefit from tissue collections must be balanced so that the research priorities of biobanks address the heterogeneous needs of diverse human populations...

exploited? Additionally, how can researchers ensure that benefits are shared across national boundaries?

Advances in IT platforms will undoubtedly go some way to addressing the ethical and practical challenges associated with biobank networks. Work done at Oxford University as part of the EnCoRe (Ensuring Consent and Revocation) Project (<http://tinyurl.com/jlhpb6x>), for example, has raised the possibility of more dynamic models of consent and communication that would enable donors to direct, with much greater precision and nuance, what projects they would consent to their sample and data being used in and what they, in return, would like to receive from this research and from their biobank.

While this model of dynamic consent offers considerable promise, and has already been adopted around the globe, it requires enormous capital investment and massive technological sophistication and may, at least initially, deepen the digital divide between rich and poor. Just as importantly, such advances in IT capacity leave unanswered the more fundamental questions regarding the purpose of research involving biobanks and the sharing or dissemination of the economic and health benefits that may flow from them.

Economic challenges associated with the global networking of biobanks stem from the fact that the infrastructure and human expertise required to maintain biobanks is incredibly costly, which puts a lot of pressure on traditional not-for-profit organisational and research funding models. Commercial funding is an obvious alternative, but while the commercialisation of a biobank can be a boon to the translation of basic

research to clinical application, it is a sensitive matter with regards to ownership, property rights and benefit sharing.

All of these technical, ethical and economic issues are further complicated by differing regulatory and ethical standards in different settings. These differences raise questions about what regulatory and ethical issues come into play when tissue is collected in one country and stored in another, and whether a biobank must respond to the regulatory and ethical standards and practices of the home country of the donor or the national location of the biobank.

International organisations such as the International Society for Biological and Environmental Repositories (ISBER), the United Nations Educational, Scientific and Cultural Organisation (UNESCO) and the Organisation for Economic Co-operation and Development (OECD) have put together guidelines that attempt to address the technical, legal, ethical and managerial issues relevant to collecting, storing, retrieving and distributing samples from human bodies for diagnostic, therapeutic, educational, forensic and research purposes. Currently, however, the operations of biobanks still operate according to national governance frameworks.

This will need to change if global networking of biobanks are to achieve their scientific promise. The competing demands of donors, investors, researchers, medical practitioners, patients, families and others who might benefit from tissue collections must be balanced so that the research priorities of biobanks address the heterogeneous needs of diverse human populations in meaningful and ethical ways.

Global biobanks are set to provide revolutionary insights into the causes of morbidity and mortality, and lead to substantial transformations in the delivery of personalised medicine. However, the potential benefits of “going global” will be undermined if technical, ethical and economic considerations are not systematically considered and resolved.

Paul H. Mason, Wendy Lipworth and Ian Kerridge research the ethics of global biobanks at the Centre for Values, Ethics and the Law in Medicine at The University of Sydney.