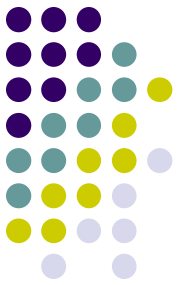
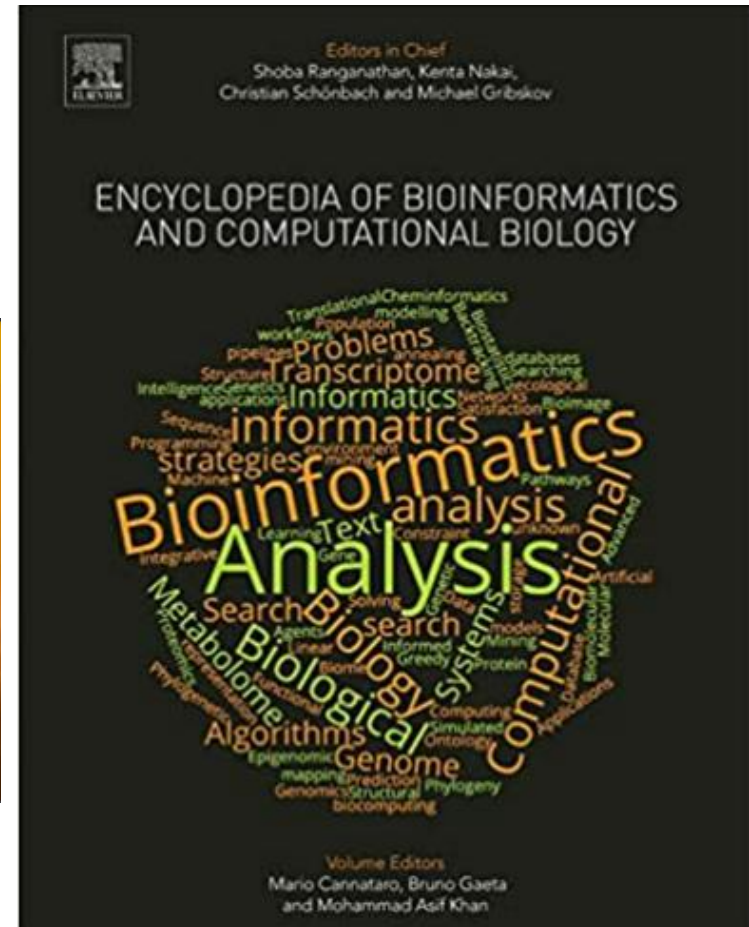
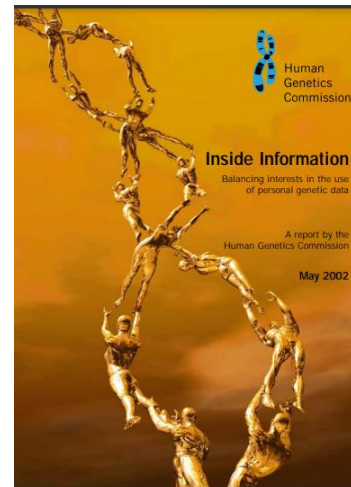
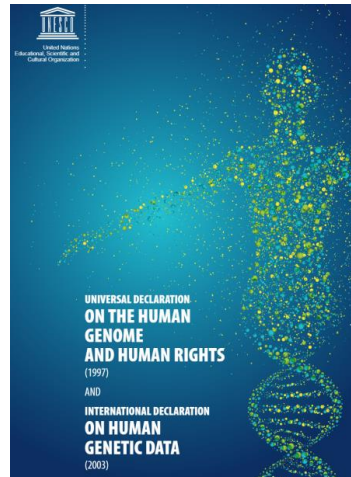
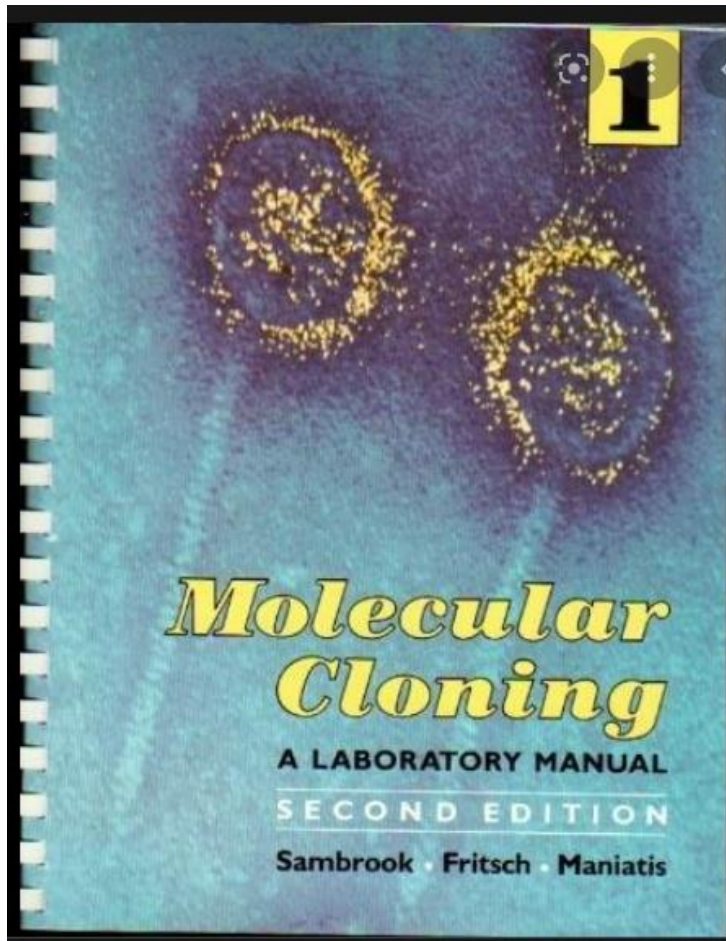


ELSI in Human Genomics and Data Sharing

Perspectives & The Pan Asian SNP Consortium Experience

Maude E Phipps, PhD.
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School of Medicine and Health Sciences
Monash University (Sunway Campus)
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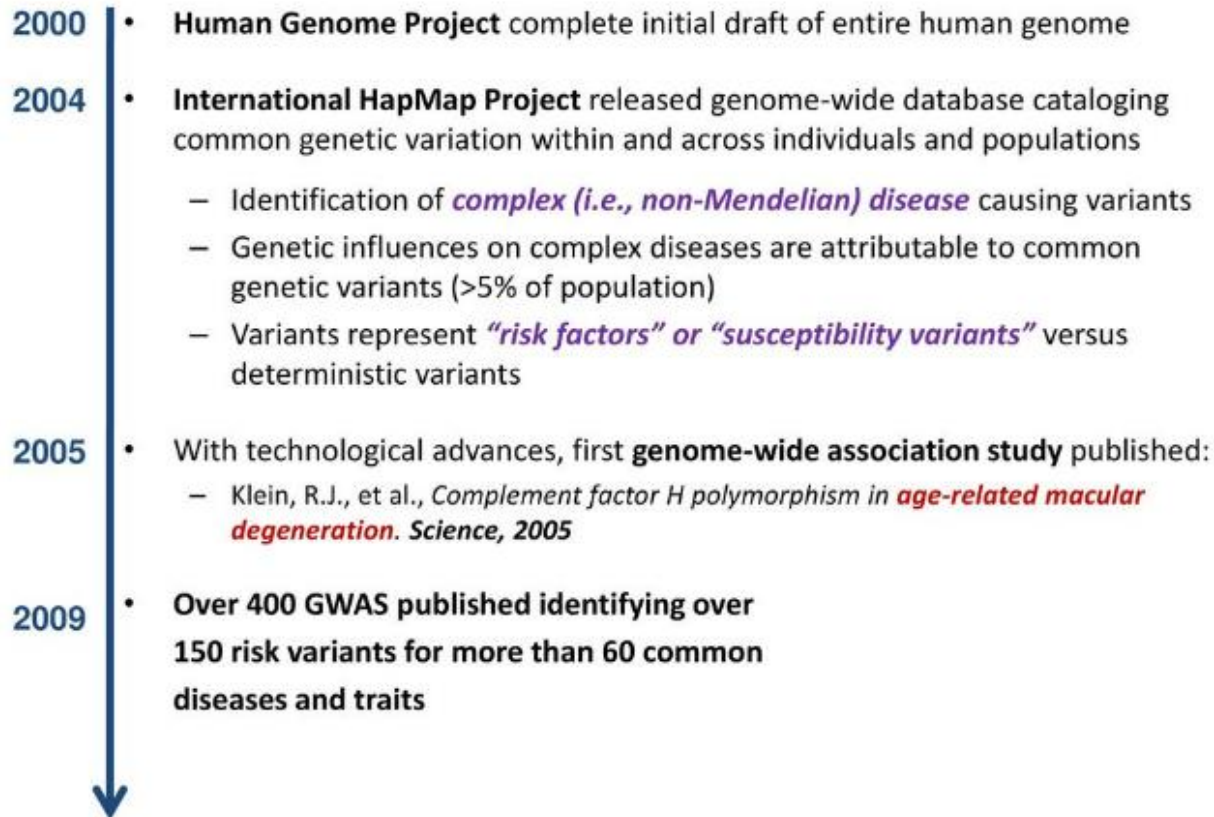


Why bother with 'ELSI' in genetics?



- Historical aspects & exceptionalism
- Ethical and Social concerns arising from current genetic research and databases
- Select 'Focus Areas'
- The PANSP experience

Human Genomics over the past 20 years



Human Genomic Databases - Online repositories of genomic variants, mainly described for a single or more genes or specifically for a population or ethnic group, aiming to facilitate diagnosis at the DNA level and to correlate genomic variants with specific phenotypic patterns and clinical features.

Today.....



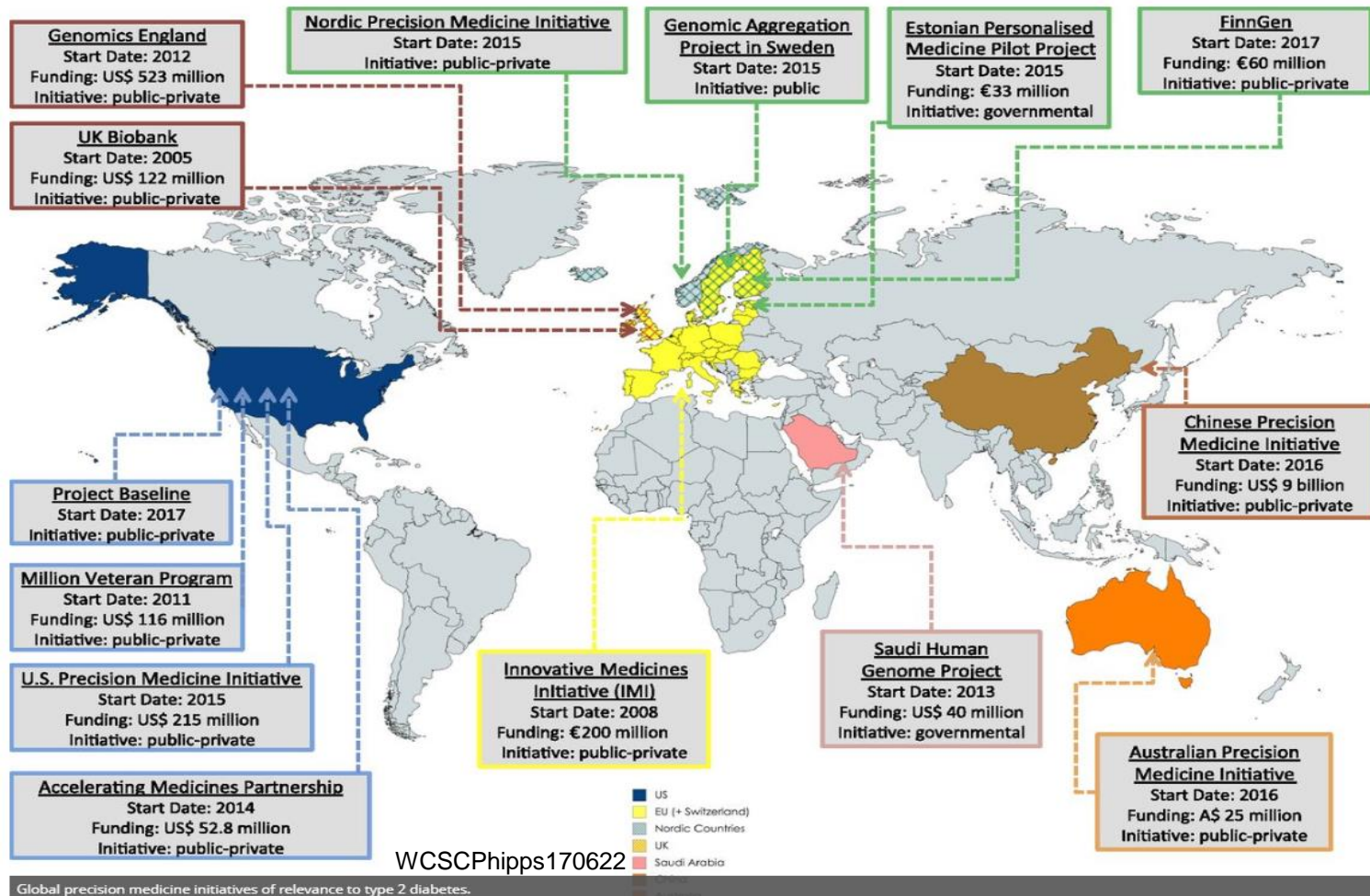
genomics databases list

[All](#)
[Images](#)
[News](#)
[Videos](#)
[Maps](#)
[More](#)

About 20,000,000 results (0.53 seconds)

Genomic Databases

- Database of Genomic Str
- Database of Genotypes &
- Database of Single Nucle
- GenBank. ...
- Gene. ...
- Gene Expression Omnib
- Gene Expression Omnib
- Genome Data Viewer (G)



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Global precision medicine initiatives of relevance to type 2 diabetes.

Genetic information is Unique

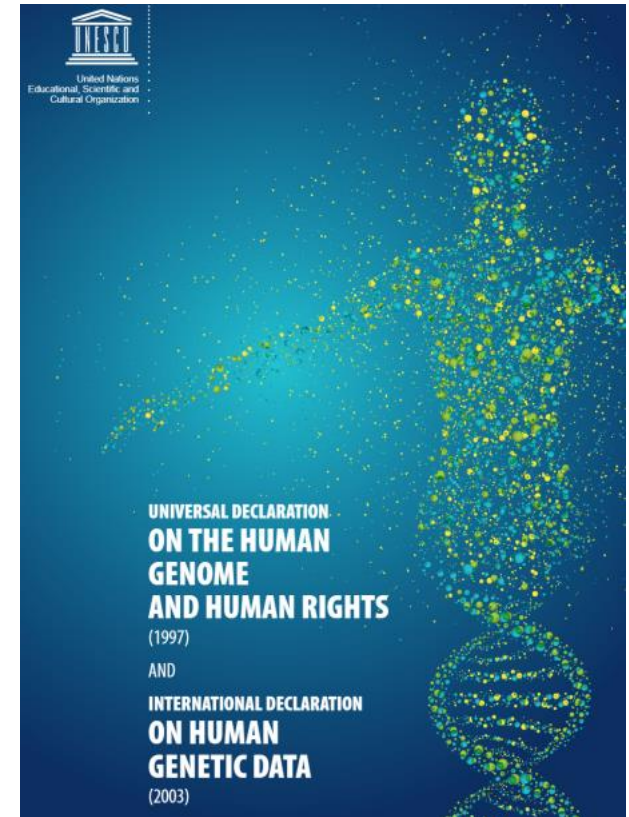


UNESCO regards the human genome as the “heritage of humanity and believes it should be protected and passed on to future generations and that advances in science need to be considered in the light of human rights.

The implementation of the Declaration is urgent since scientific progress is accelerating and both giving humankind hope and creating ethical dilemmas.

These Guidelines seek to identify tasks devolving on the different actors in the implementation of the Declaration and modalities of action for their achievement.

1) What to do 2) How ? 3) For Who ?



Genetic information is unique



Article 4 – Special status

- (a) Human genetic data have a special status because:
 - (i) they can be predictive of genetic predispositions concerning individuals;
 - (ii) they may have a significant impact on the family, including offspring, extending over generations, and in some instances on the whole group to which the person concerned belongs;
 - (iii) they may contain information the significance of which is not necessarily known at the time of the collection of the biological samples;
 - (iv) they may have cultural significance for persons or groups.



ELSI focus areas

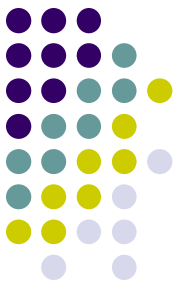
- Autonomy
 - Privacy, Impact on Communities
- Beneficence
 - Benefits and Data Sharing
 - Non discrimination
- Incidental findings

Autonomy



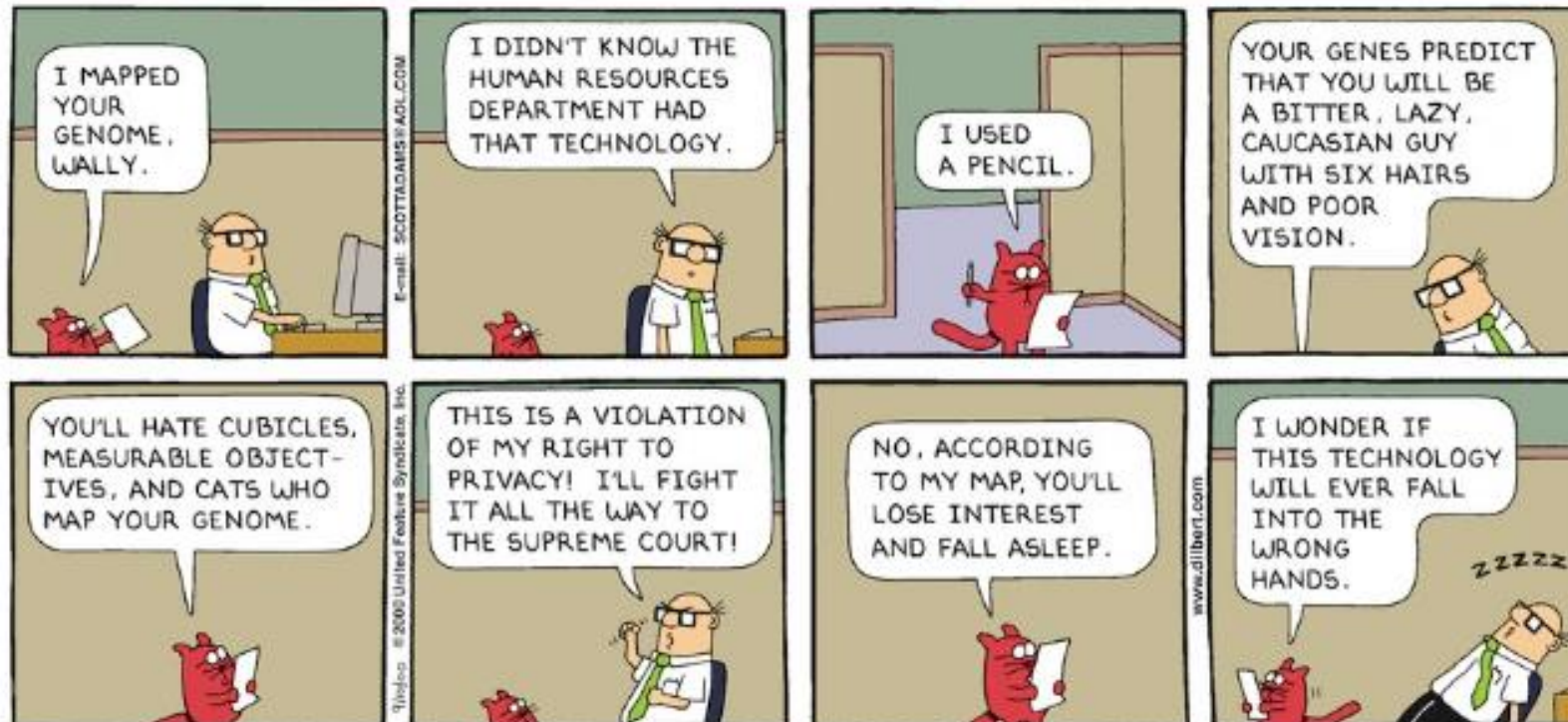
- The principle of respect for a person, to treat them as a person with their own values, reflectively held beliefs and desires.
- Dignity and worth as human beings
- Underpins importance of Informed Consent and Confidentiality for Privacy and Data sharing

Sunday September 10, 2000



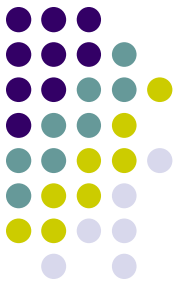
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The slippery slope of generalizing 'Informed Consent'



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HEALTH LAW
FEB 2011

Genetic Research among the Havasupai: A Cautionary Tale

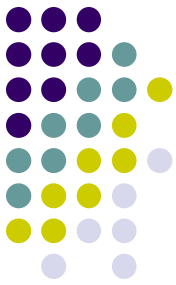
Robyn L. Sterling, JD, MPH

Citation PDF Altmetric



Imagine that you donated a bit of blood to a researcher whom you believed intended to identify a genetic link to a disease ravaging your community, only to discover years later not only that you had been misled, but that other researchers were mining your DNA for reasons that were never disclosed to you. What would you do? This was the case for the Havasupai Tribe in Arizona, who learned that researchers at Arizona State University (ASU) had gathered blood samples from them to search for a link to diabetes but used the samples to look for other diseases and genetic markers, thereby violating the basic tenets of human subject research. To determine where the breakdown between the Havasupai Tribe and ASU occurred, let's look at community-based participatory research and its underlying principles of informed consent.

“...by failing to follow proper protocols and regulations, a researcher engaging in CBPR may inflict permanent harm on the participating community and chill future research among disadvantaged populations.”



Beneficence



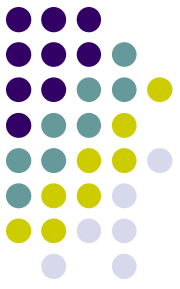
- To benefit a person and act in their best interests
- Provide a good outcome from an interaction
- Prevent harmful outcomes
- ‘Harm’ can be a matter of perspective



Who **benefits** from genomics R & D ?

- Academia
- Medical Fraternity
- Civil Society
- Governments
- Industry
- Participants / Communities

Non discrimination



Article 7 – Non-discrimination and non-stigmatization

- (a) Every effort should be made to ensure that human genetic data and human proteomic data are not used for purposes that discriminate in a way that is intended to infringe, or has the effect of infringing human rights, fundamental freedoms or human dignity of an individual or for purposes that lead to the stigmatization of an individual, a family, a group or communities.
- (b) In this regard, appropriate attention should be paid to the findings of population-based genetic studies and behavioural genetic studies and their interpretations.

Education



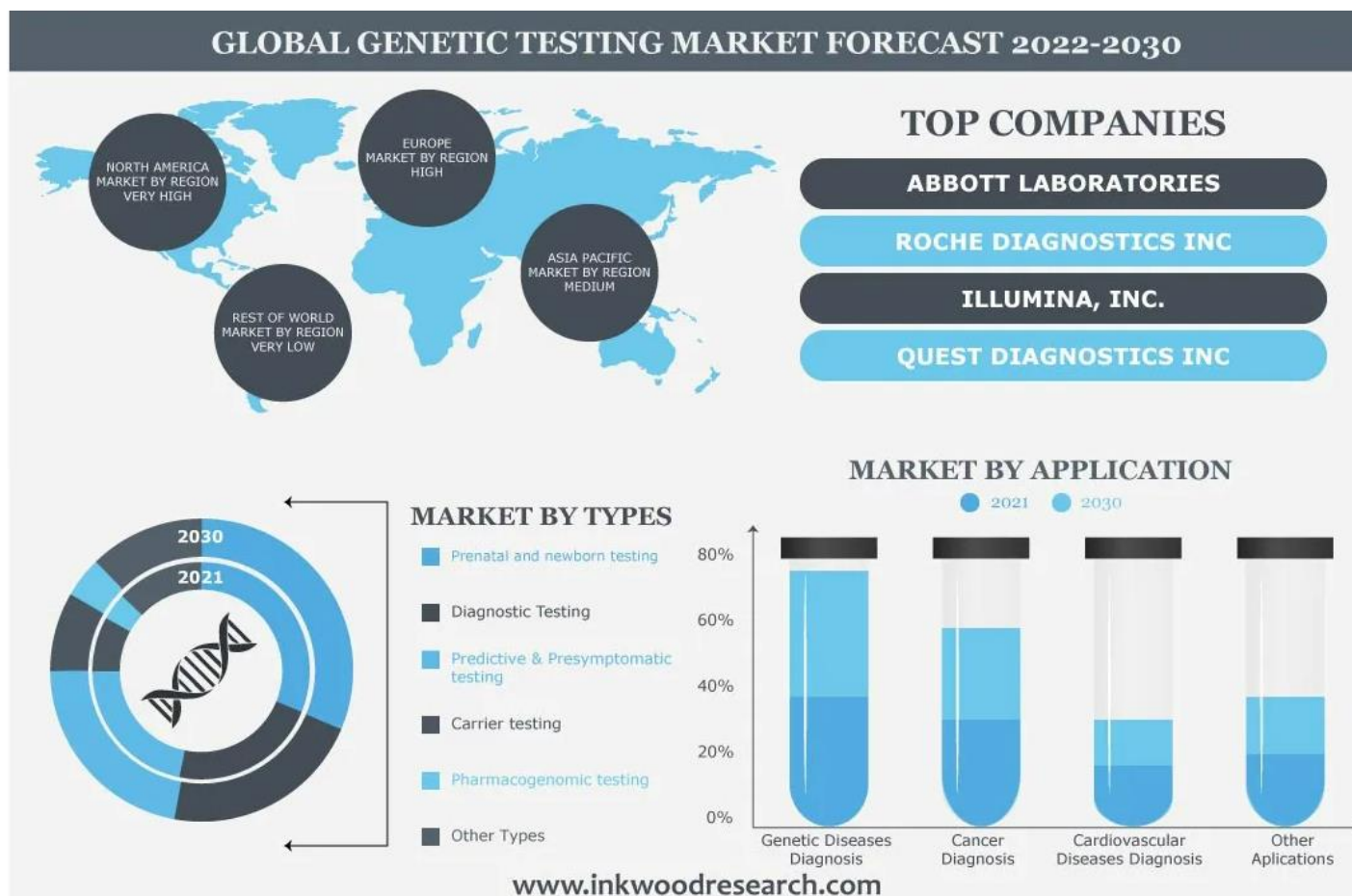
Employment



Insurance



Genetic tests and Incidental findings



According to a new [market research](#) report launched by Inkwood [Research](#), the Global Genetic Testing Market is set to reach **\$27.81 billion by 2030**, recording a CAGR of 10.25% during the forecast period, 2022-2030.



Incidental findings

- When the investigation of an individual's DNA sequence for one purpose, results in **discovering something important that was not part of their original aim** but might have **clinical/serious significance**.
- e.g. During a study on intelligence, a participant is discovered to have a Huntington's disease mutation, an incurable neuromuscular disease that eventually causes death, later in life.

What do you do ?

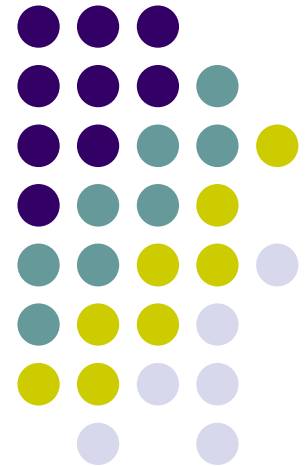
According to the National Human Genome Research Institute, a researcher should report incidental findings if those findings meet 3 categories: 1) findings can be validated, 2) findings have health implications, and 3) knowing the findings will be helpful in taking specific actions to improve the participant's health.

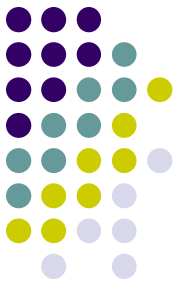
How do you/does your institution deal with this?

Policy Review Board

Regulation from Within

Pan Asian Single Nucleotide Polymorphism
(PASNP) Initiative Experience
(2005-2010)





- Investigation of 1928 individuals representing 73 Asian populations + 2 non Asian HapMap populations



Some Indigenous communities I worked with



Proto-Malay

- Austronesian
- Seafarer & Farmer
- Sub-tribes/Locations: Orang Seletar (Johor, Near Singapore), Jakun (Tasik Chini), Temuan (Jejebu)



MEP2049

15/12/2009

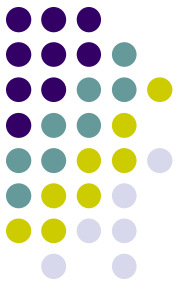


Source: JHEOA (2008)

PRB Vision Statement ?



- “What we’re doing must be scientifically sound, in line with technological advances involve international collaborative efforts and ethically justifiable in our quest for greater knowledge and wisdom of the human genome, evolution and health”



PRB - Terms of Reference

- Identification of relevant ELSI parameters
- Consultation, engagement and discussion (bioethicists, researchers, policy makers, etc, within HUGO, other agencies and stakeholders)
- Policy formulation and periodic review for the duration of the initiative
- Collation of documentary evidence – Informed consent, approvals of IRBs, other regulatory authorities .
- Addressing of ethical concerns relevant to research findings, publications and implications, especially in the Asia Pacific

Bioethics Instruments / Guidelines applicable to the PRB-PASNPI



- International
 - UNESCO
 - Universal Declaration on Bioethics and Human Rights
 - Universal Declaration on the Human Genome and Human Rights (2000)
 - Bioethics Committee at Work – Procedures and Policies (2005)
 - WHO
 - Genomics and World Health (2002) Summary and Guidelines
 - Adv. Comm. Health Res. - T. Pang, D. Weatherall, D. Brock and H.L.Chee
- National / Institutional
 - National Regulatory Frameworks, Organizations, IRBs, etc and Policies therein

International Articles relevant to Human Genome research



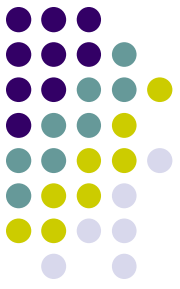
- Article 10 (Informed Consent, IRB)
 - No research or research applications concerning the human genome, in particularly the fields of biology, genetics and medicine, should prevail over respect for human rights, fundamental freedoms and human dignity or where applicable of groups of people.
- Article 12 (Research and Benefits Sharing)
 - Benefits from advances in biol, gen., med., shall be made available to all.....
 - Freedom of research, progress of knowledge ... freedom of thought. Appl. of res...human genome... shall seek to offer relief from suffering and improve the helath of individuals and humankind as a whole.

Intl. Articles relevant to Solidarity and Intl. Cooperation



- Articles 17, 18, 19
 - refer to intl. scientific, cultural cooperation, genetically based and genetically influenced diseases, human diversity and genetic research, industrial and developing countries.
- Existing scientific collaborations among investigators and institutions
- Technology transfer and Sustainability
- Benefits Sharing

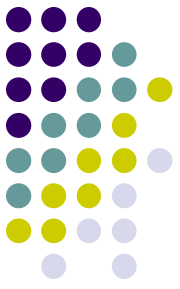
PRB – Bioethics indicators



- **IRB approvals*** (each investigator / institution)
 - Mandatory including Primary approvals from relevant national ministries and agencies
 - Copy of the IRB approval reference code / number kept at the Secretariat in PRB records (for each researcher /group)
- **Informed consent*** (volunteers & archived samples)
 - Mandatory and responsibility of each investigator/group.
 - A copy/copies of a signed and verified IC form to be submitted for PRB records
 - Other mechs. utilized

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PRB – Bioethics indicators (for disc.)



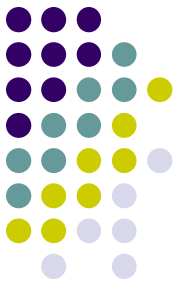
- ‘Conflict of Interest’ issues
 - Public fund utilization and accountability
 - Translation into research / training / tech transfer / capacity building / intl. cooperation, other output
 - Public and institutional database access and utility
 - Private sector partnerships and developments
 - PANSPI : Public Inst. + Affymetrix
 - Other partners
 - IPR, patents, commercial spin-offs / activities
 - NOT APPLICABLE

PRB – Bioethics indicators (for disc.)



- Benefits Sharing
 - Community outreach in developing countries / populations
 - Mechanisms for immediate benefits to participants
 - Longer term benefits to be discussed
 - Sharing of results and findings with participants
 - Global benefits sharing agreements
 - Tangibles
 - Intangibles

Sunday July 09, 2000



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BY
SCOTT ADAMS

MY SOFTWARE WILL
CREATE HUMAN
SIMULATIONS FROM
DNA SAMPLES.

WHAT'S THE
MARKET
APPLICATION?

WELL... THERE ARE
MANY VARIOUS
APPLICATIONS.

NAME
ONE.

WELL... SOMEDAY
THE ENTIRE
HUMAN GENOME
WILL BE MAPPED
AND DECODED.

YOU COULD TAKE A
HAIR SAMPLE FROM
A WOMAN WHO
REFUSES TO DATE
YOU...

AND CREATE A SOFT-
WARE SIMULATION
OF HER TO KEEP IN
YOUR COMPUTER
WATCH.

YOU COULD HAVE
ONE BUTTON TO
FEED HER AND
ONE BUTTON TO
PUNISH HER.

I'D
BUY
IT.

CAN YOU
ADD A
BUTTON?

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Your thoughts.....

- 1) How does personal genetic information affect and individual and society's perception of that individual
- 2) Do healthcare personnel properly counsel parents about the risks and limitation of newer genetic technologies?
- 3) Should testing be performed when no treatment is available ?
- 4) How do we as a society balance current scientific limitations and social risks for long term benefits ?



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