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BIOL3120 – Human Genetics and Evolutionary Medicine

Ethics in Human Genetics

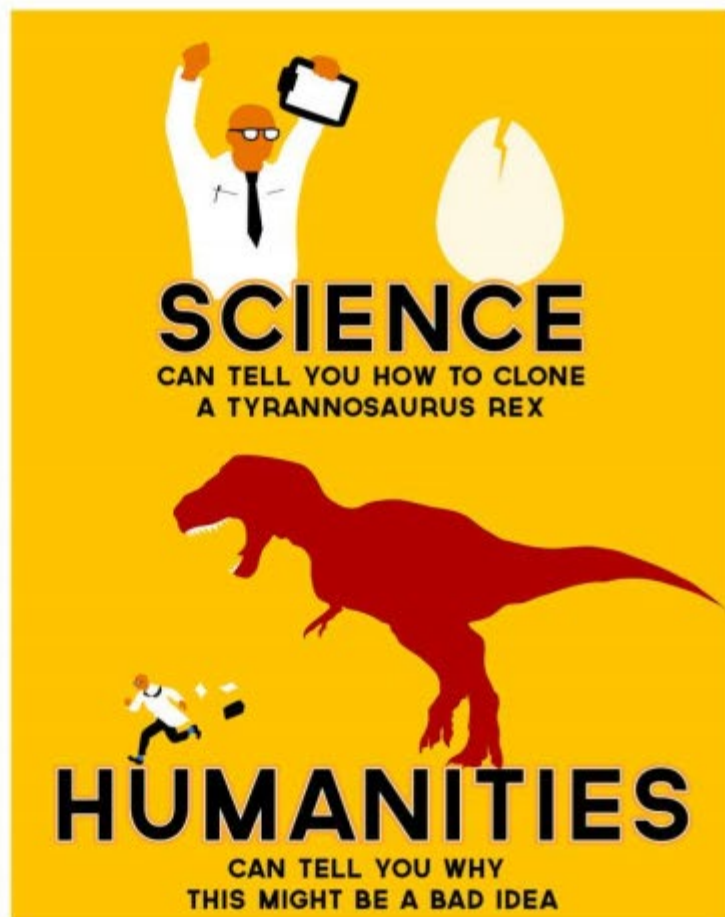


Foundations in Ethics and Society

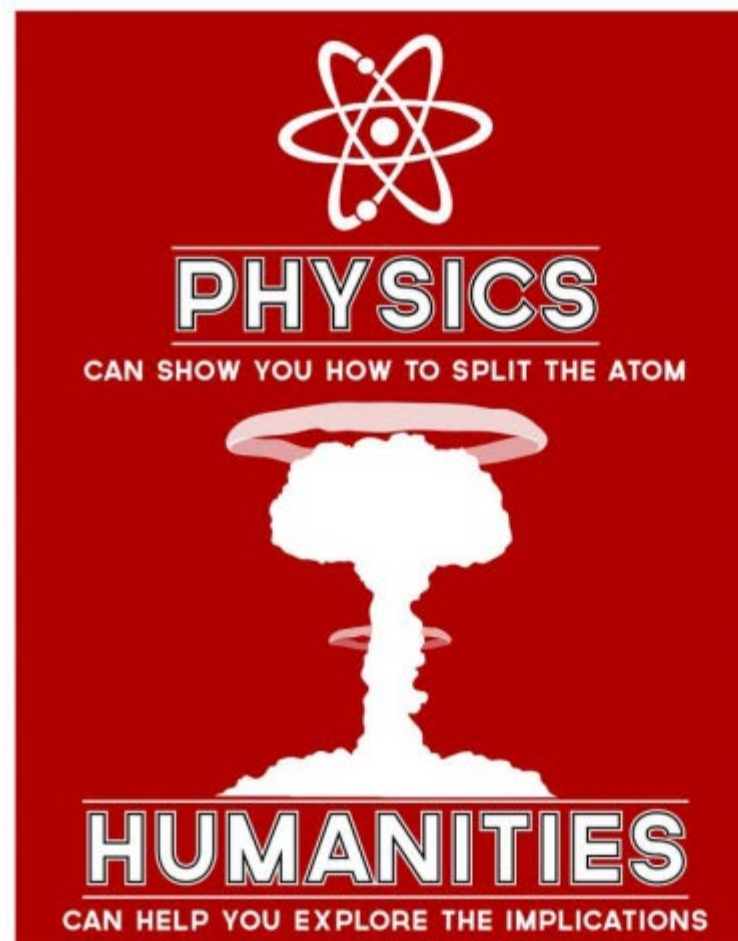
BLENDING SCIENCE AND HUMANITIES



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Human Genetics

With great power comes great responsibility

- Human Genome Project
- Next Generation Sequencing
- Genetic Testing
- Pre-implantation screening
- Genome Wide Associated Studies
- Personalised Medicine
- Genome Editing
- 3 parent children



BIOL3120 –Ethics in Human Genetics

LEARNING OBJECTIVES



On successful completion of this lecture, you will be able to:

- Identify the principles of bioethics
- Identify ethical dilemmas in genetics
- Practice ethical decision making



The Georgetown Principles of Bioethics

Bioethics –what is it?



The ethical evaluation

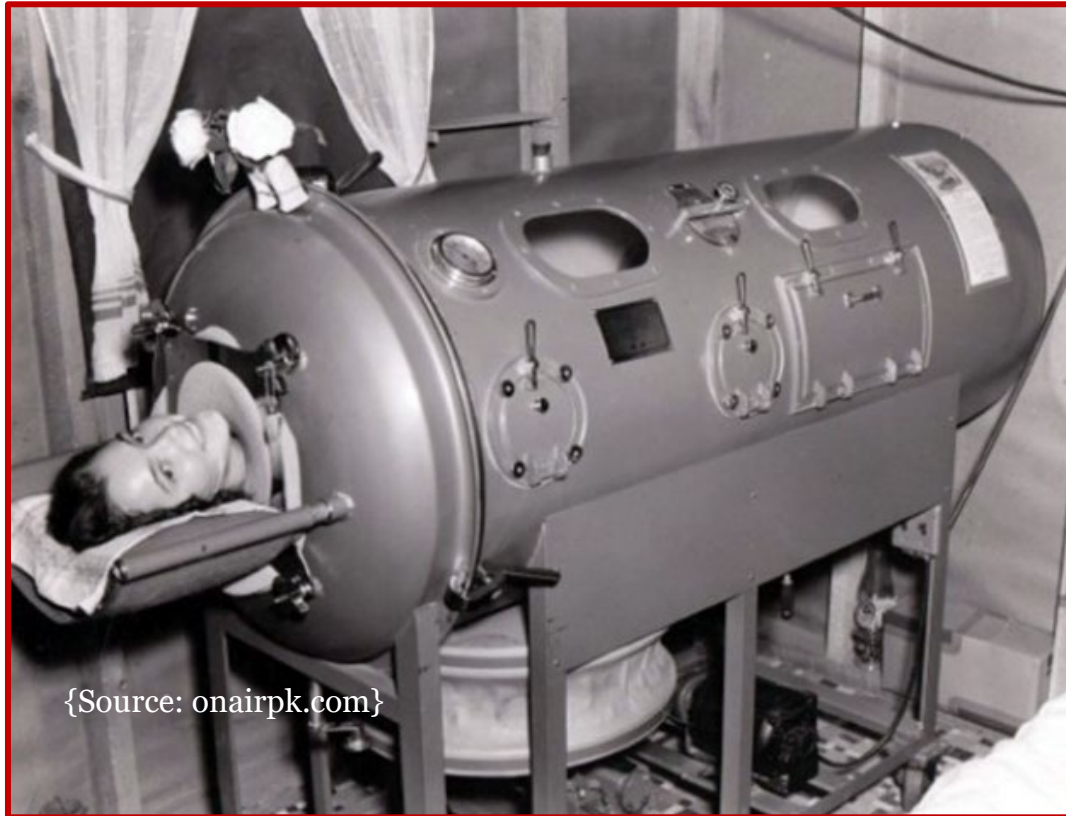
of moral dilemmas

affecting human beings

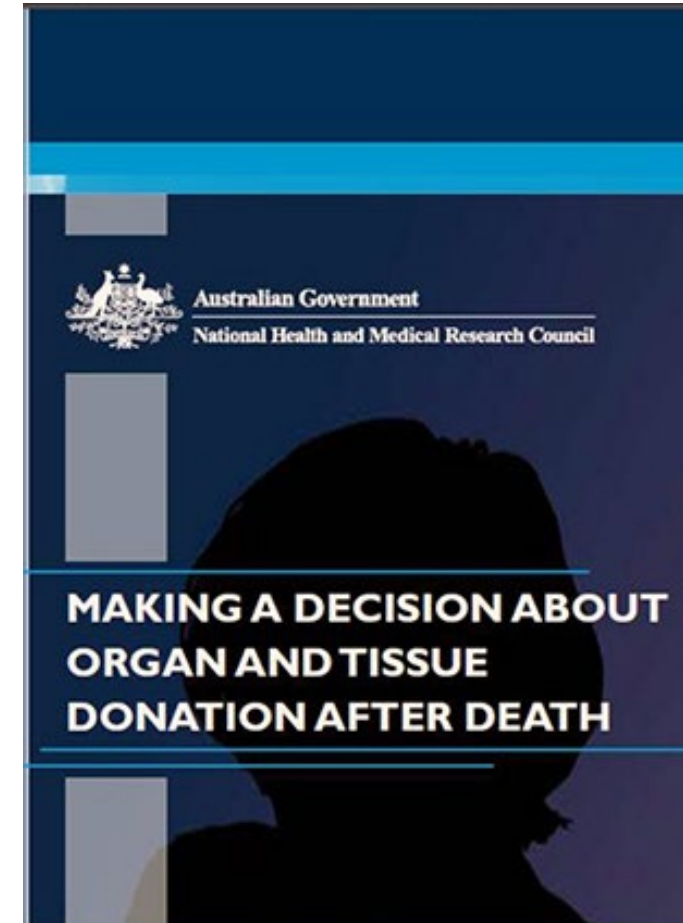
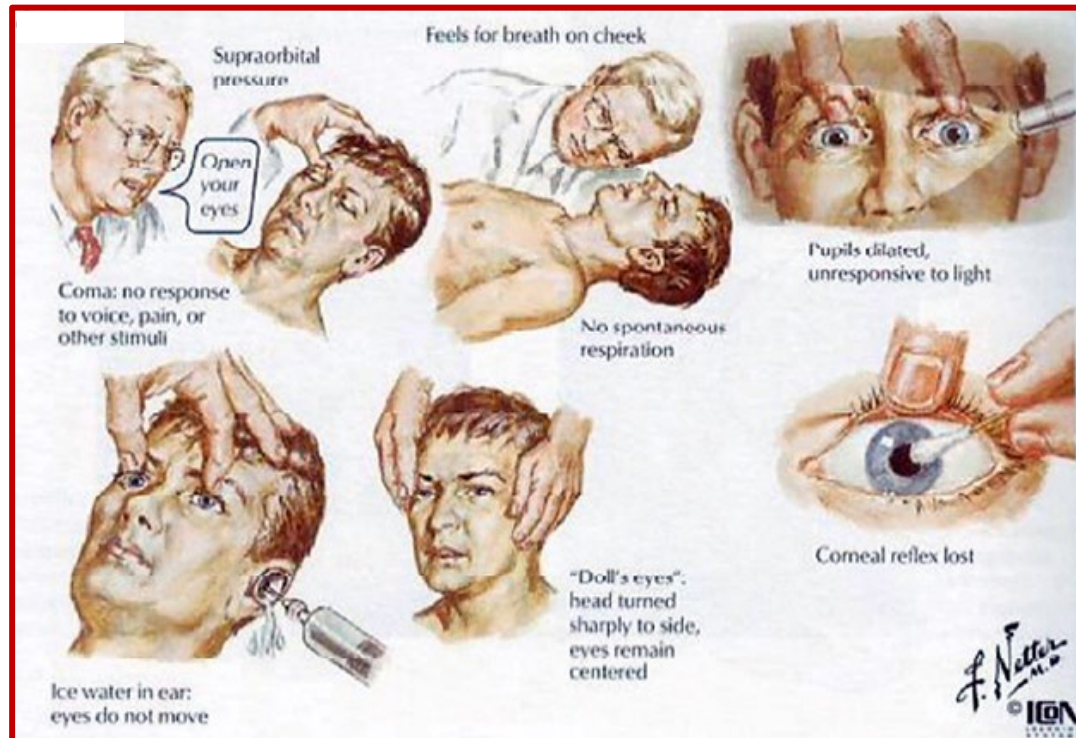
in medicalised settings.

- technological advances
- societal influences

Technological Advances - Ventilation



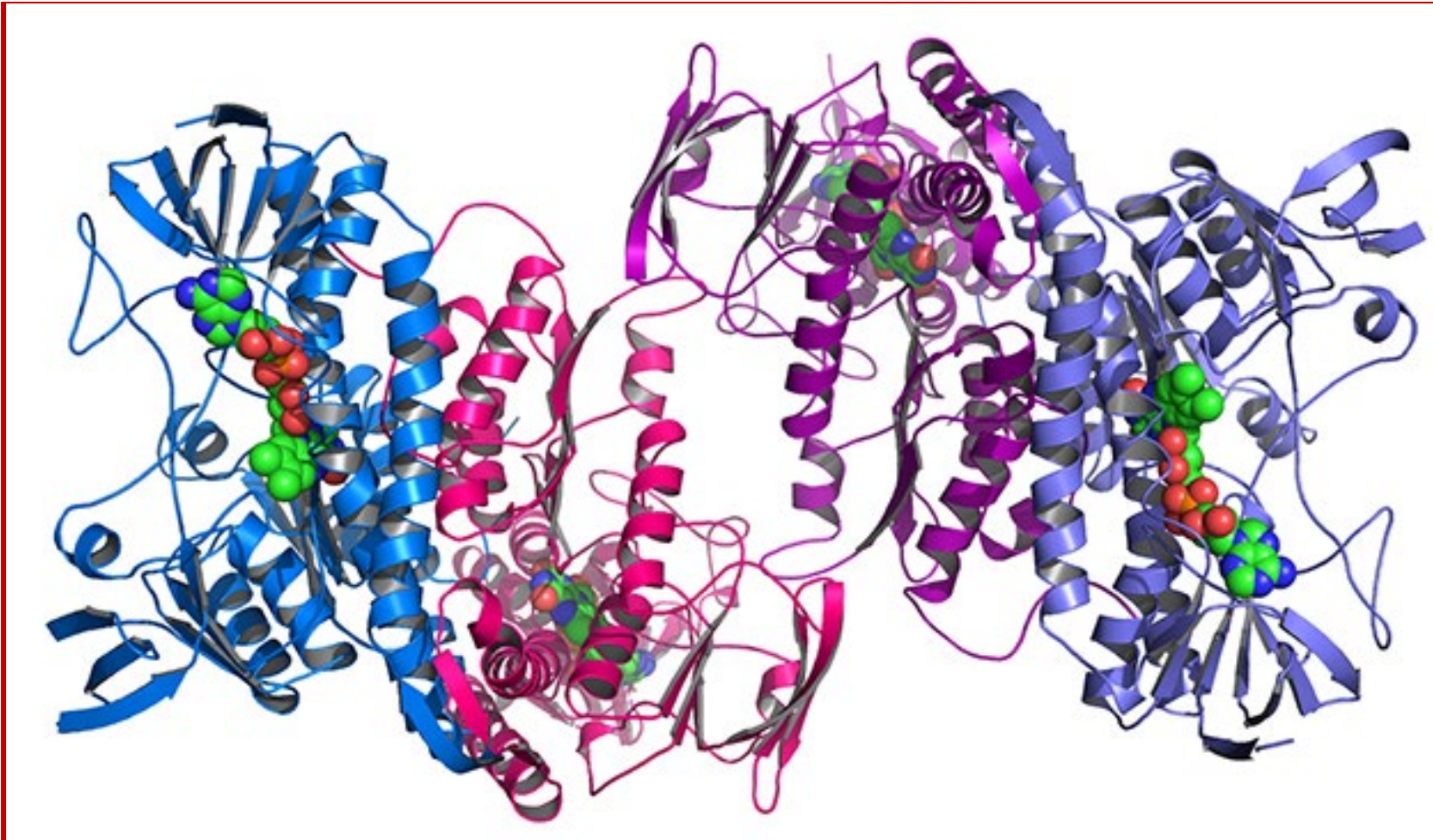
Technological Advances - Transplantation



Technological Advances – Artificial Reproductive Technologies



Genetics, Genomics and Proteomics



{Source: Wiki Commons}

Bioethics Institutions (USA)



The Hastings Center {1969}



The Kennedy Institute of Ethics {1971}

The Principles of Bio-Medical Ethics

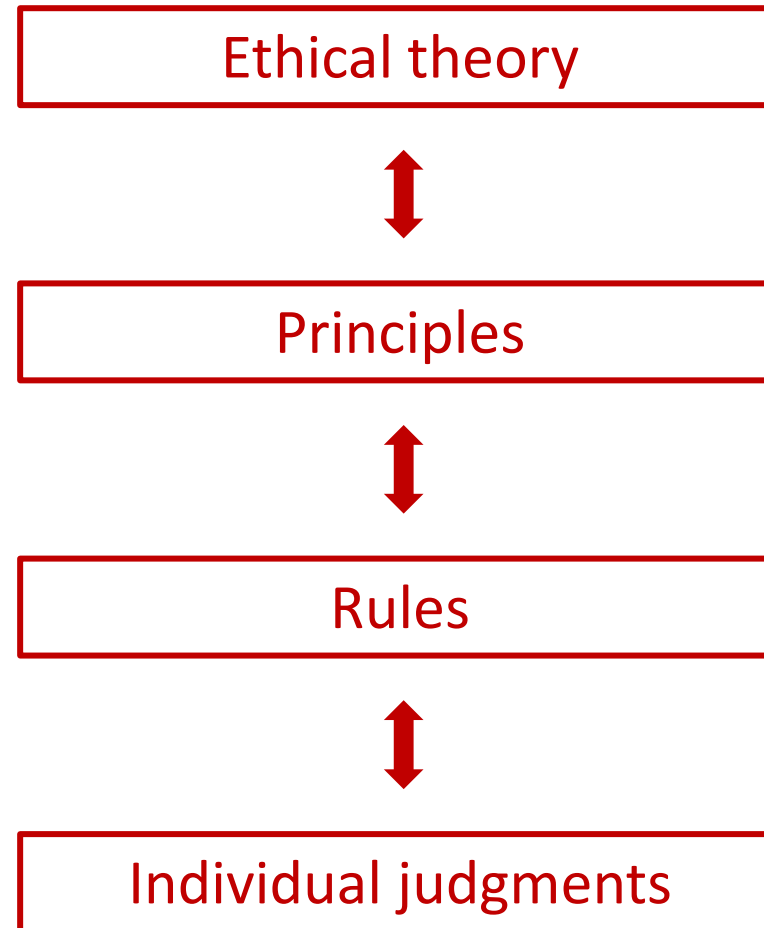


Tom Beauchamp

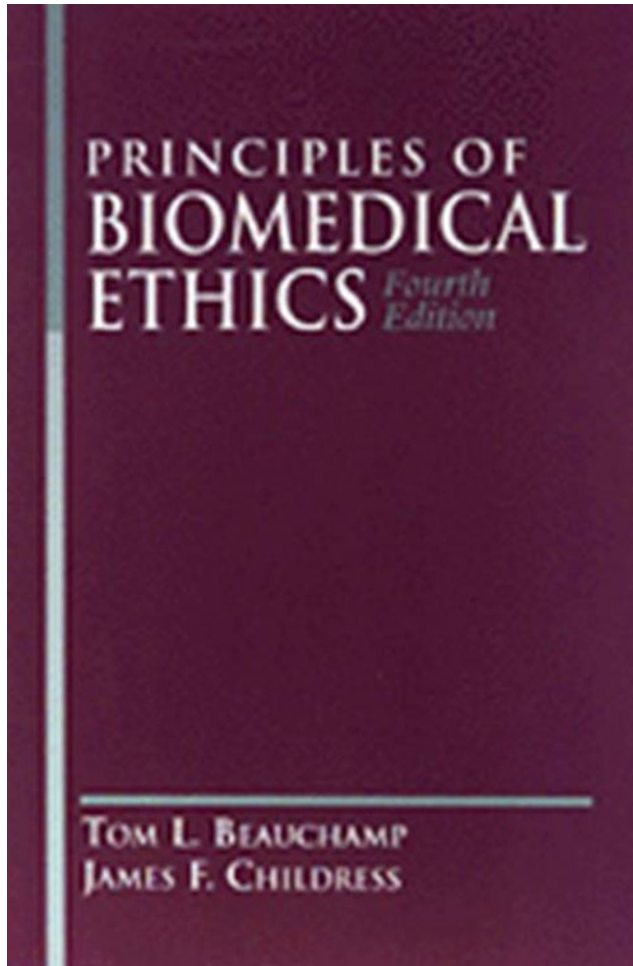


James Childress

A Hierarchy



Principles of Bioethics



Respect for Autonomy

Non-Maleficence

Beneficence

Justice

The Practical Impact of Bioethics

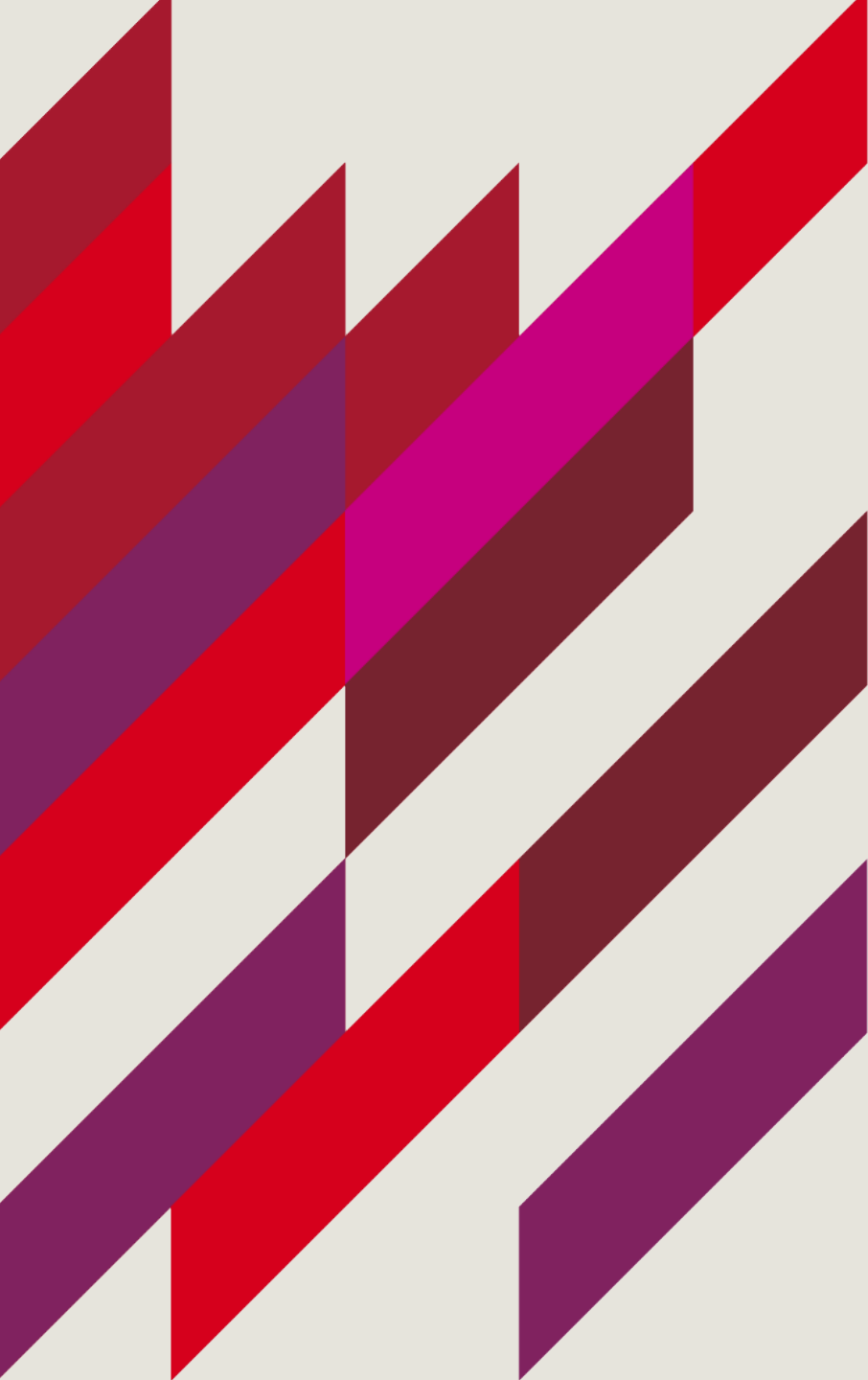


A third voice (beyond scientists and politicians) in the assessment of the impact science and technology

Brought new minds and expertise into play

👉 Civic management through guidelines and regulations

Civic oversight of biomedical research



Ethical Dilemmas in Genetics

Genetic exceptionalism

- “**Genetic exceptionalism** is the belief that genetic information is special and so must be treated differently from other types of medical data or other personally identifiable information.”
- Some genetic information could cause discrimination
 - Job?
 - Insurance?
 - College scholarship?
- Genetic Information Nondiscrimination Act (GINA) 2009
 - illegal for health insurance providers to use or require genetic information to make decisions about a person's insurance eligibility or coverage (health insurance only)
 - illegal for employers to use a person's genetic information when making decisions about hiring, promotion, and several other terms of employment

Disclosing to family members

- Telling other family members about genetic finding
- Family cut off (separation, repartnering, etc) or never a relationship?
 - Often can be related to symptoms of condition
- Unknown paternity / adoption?
- Condition details? Lead to treatment/management?

Disclosing to family members

- Allows for the use or disclosure of genetic information to a patient's genetic relatives

“If the organisation reasonably believes that this is necessary to lessen or prevent a serious threat to life, health or safety of the relative, even if the threat is not imminent and consent has not been given.”

Privacy Legislation Amendment Act 2006

Disclosing to family members

- **Only applies to Private practice**
- Allows for disclosure and use of information without consent, provided that such disclosure is:
“Necessary to lessen or prevent a serious threat to the life, health or safety (whether or not the threat is imminent) of an individual who is a genetic relative of the individual to whom the genetic information relates”
- A medical practitioner must authorise disclosure and there must be consultation with appropriate colleagues
- Not an obligation – no duty to warn

Potential benefits of patient disclosing

- Clarification of the risk status of clinically unaffected relatives
- Possibility for steps to be taken to reduce the risk of disease or allow early diagnosis and management
- Avoidance of the need for other investigative interventions if genetic testing identifies the relative as a non-carrier.
- Avoidance of mistaken diagnosis (of another condition) and inaccurate treatment.
- Shared knowledge of the genetic condition within the family may help to avoid family breakdown and anger.

Potential risks of disclosing without consent

- Possibility of the privacy of the patient being affected.
- Possibility of losing the patient's trust and confidence.
- Difficulties in the process of advising genetic relatives even if patient is willing to share genetic information.
- Potential for patient uncertainties about the practicalities of disclosure.
- Genetic relatives feeling that receiving unsolicited information about possible genetic risk is an invasion of privacy.
- Perceived pressure on genetic relatives to undertake genetic assessment.

Potential risks of not disclosing

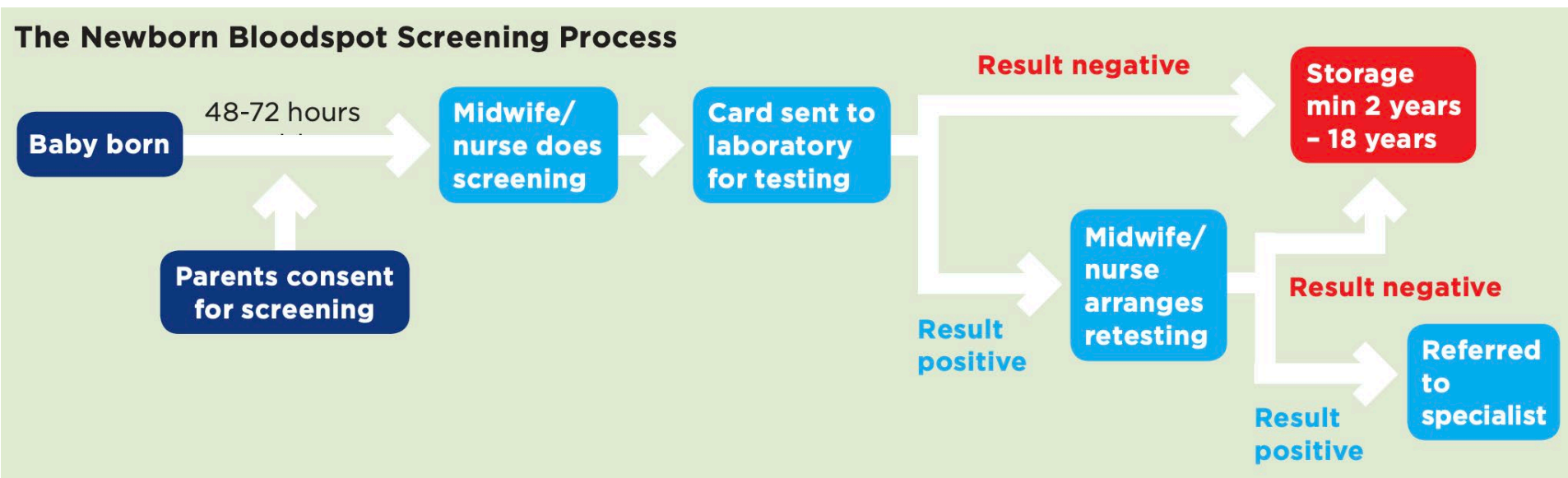
- Relatives are left unaware of potential risk.
- Potentially preventable harm cannot be averted.
- Relationships within family may be damaged when relatives discover that information was not passed on to them.
- Relatives who were not informed due to a deliberate decision by a health practitioner not to disclose may be distressed and experience harm as a result.

Genetic testing of asymptomatic children

- What is the condition?
 - Treatment / management options available?
- Loss of autonomy?
 - To child? Parents' autonomy?
 - Age/maturity of child?
- Beneficence – what are the potential outcomes?
 - What are the best interests of those involved?
- Non-maleficence
 - Net benefit?
 - What is harm?

Newborn screening

- ~100,000 babies tested per year in NSW
- 25 conditions tested for which early intervention greatly increases health
- ~110 babies diagnosed through this process per year



Newborn screening

- Storage of the sample
 - Permission to store required? For how long? **Real consent?**
 - Card destroyed when child turns 18 since they didn't consent – parents did
 - Should the samples be able to be used? Under what conditions? - Who should be able to access samples?
 - Sometimes used for forensic purposes, e.g. Post-mortem identification
- Whole genome sequencing of the newborn sample - Trials in the UK and the US
- Data storage/security?

He Jiankui – CRISPR babies

- Fertility service for HIV positive fathers, HIV negative mothers
- IVF with CRISPR used to modify CCR5, attempting to confer HIV resistance
- Alarm raised in November 2018, prompted announcement of twin birth in October 2018, third baby born 2019.
- 2019 Sentenced to 3 years prison, fined \$430,000 , barred from reproductive tech work for life. Had falsified regulatory paperwork





Ethical Decision Making

Principles of bioethics in medical practice and research

- **Autonomy:** respect autonomy - educate, communicate, consult, respect and empower.
- **Justice:** Promote fair distribution of resources, respect for rights and respect for morally acceptable laws.
- **Beneficence:** provide net benefits
- **Non-maleficence:** do no avoidable harm, to individuals or groups.
- Reflected throughout medical/research policies etc.

Autonomy

- The principle of autonomy and respect for the dignity of each human being
- Most commonly reflected in the requirement that a person must give his or her consent prior to any treatment or testing, and that such consent be informed by an understanding of adequate information.
- In relation to genetic information, there may be a need to extend the concept of informed consent to provide a similar degree of respect to a family or even a broader community.
- Confidentiality
- See: [consent form](#)

Consent (part of autonomy)

- “Autonomous authorisation of a medical intervention ..by individual patients” (Beauchamp and Faden 2004)
- Most visible way the health system manifests respect for patient autonomy
- Ensuring competent patients are able to make autonomous decisions
- Non-competent patients are protected from harm

Beauchamp T and Faden R (2004) Informed consent: II Meaning and elements of informed consent. Encyclopaedia of Bioethics (3rd ed). S Post. New York, MacMillan Reference, USA

Justice

“treatment be made available on a fair and equitable basis.”

- How genetic information can be used so as to avoid unfair (or unlawful) discrimination.
- Access to genetic testing
- As a matter of social justice, access to genetic testing services which have been shown to have potential to provide significant health benefits should not be dependent on where a person lives or on their socio-economic status.

Beneficence

“health care be aimed to produce beneficial outcomes”

- Aim for actions which promote the well-being of others, taking into account the best interests of patients and their families
- This principle would be relevant to:
 - Is it beneficial to conduct genetic tests on families or communities (especially where there is no known treatment)?

Non-maleficence

- Most commonly reflected in the requirement that, on balance, treatment should not cause harm.
- This principle would be relevant to the debate about conducting genetic tests on children who have not shown any symptoms but might be emotionally and intellectually harmed by the test results.
- Net benefit over harm
- May have no obligation to benefit but do have obligation not harm - informed consent for prenatal diagnostic procedures
- Assessment of individual benefit/harm balance may vary - preventive mastectomy and/or oophorectomy

Privacy and confidentiality

- The right to be let alone
- The right to personal space or autonomy
- The right of people to exercise control over their personal information or the degree of interference with their personal life
- A popular reaction to the spread of new technologies
- Simply fair information practices.

PRACTICAL ETHICS ANALYSIS FRAMEWORK

Based on:

- Rogers and Braunack-Mayer 2008. *Practical Ethics for General Practice*, 2nd edition. Oxford University Press, p 16, and
- Ethox Approach Flow Chart (http://www.ukcen.net/education_resources).

1. Summary of the issue or case

What are the facts or context that are relevant to the issue? Include, where necessary, a summary of any affected patient's history

2. Decision making process

Who is responsible for this decision?

When does a decision have to be made? E.g. End of life care / time critical

Who should be involved in this decision?

What are the procedural rules?

3. What are relevant ethical (and legal, social) questions/problems to be resolved?

Ethical issues: eg What are the medical needs of the patient and how do these compare with her preferred course of action?

Legal issues: eg What are the statutory obligations of healthcare professionals in this situation?

Social issues: eg How do differences in cultural views on breaking bad news affect the situation?

4. Identifying and analysing the options

Identify and list potential options

Look at the moral arguments for and against each of the identified options, taking the following perspectives into account:

Individual's perspective (may be patients, or citizens)

How does this option align with the individuals' needs, wants, rights, cultural issues, medical and non-medical, relationships etc?

Healthcare practitioner's duties and obligations

How does this option align with ethical duties (e.g. beneficence, trustworthiness, honesty, respect)?

What are the relevant legal & professional obligations?

Potential consequences for all stakeholders

Patient / Citizens / Practitioners / Health care team / Hospital

Community – what are the implications if this option became a general rule in similar future cases?

5. What is your recommended option and why?

Based on the significant features of this case and options presented, evaluate your option. Identify the strongest counter argument to this option and ensure you can rebut this.

6. Conclusion

A brief statement justifying your preferred course of action in relation to the ethical issues.

7. References

These should include only material that you have cited in the text. Use up to 6 recent and relevant journal articles or book chapters.

How to answer?

Ethical Dilemma:

Central Issue: What is the over-riding ethical dilemma?

Competing Considerations: What other aspects should be taken into account?

Decision making process: Which principle(s) of bioethics did you apply and what was the decision?

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