

Emerging prenatal genetic testing technologies: ethical issues and the need for empirical research

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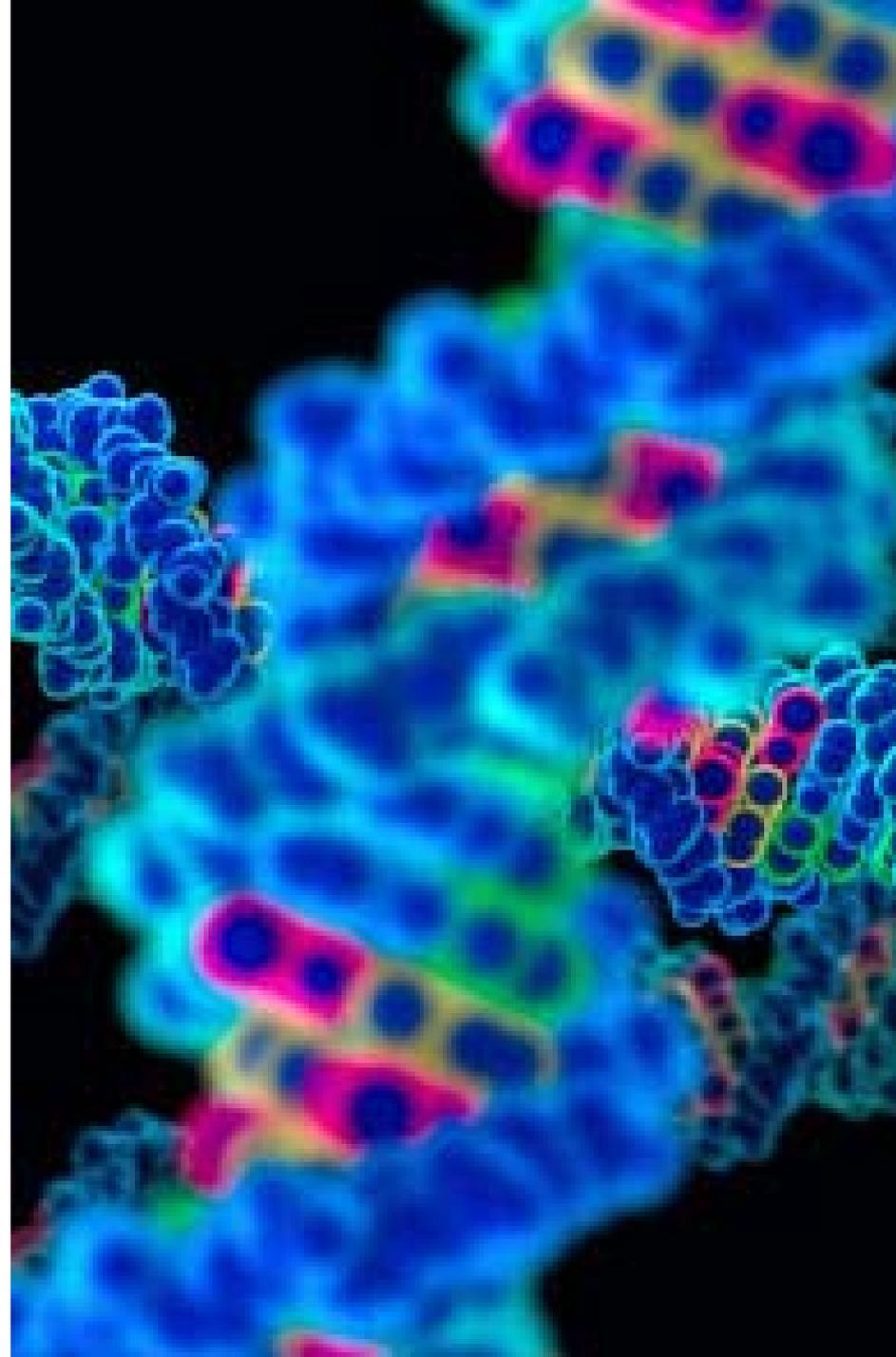
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Outline of the presentation

- Overview of NIPD technology and its applications**
- Practical issues**
- Ethical issues raised**
- Further research**



Scientific background

Cell free fetal DNA -
increases with gestation,
rapidly cleared after birth

Non invasive: simple
blood test

Diagnostic: highly
accurate results

Early: testing from 7
weeks gestation (first
trimester)



Recent developments and current use

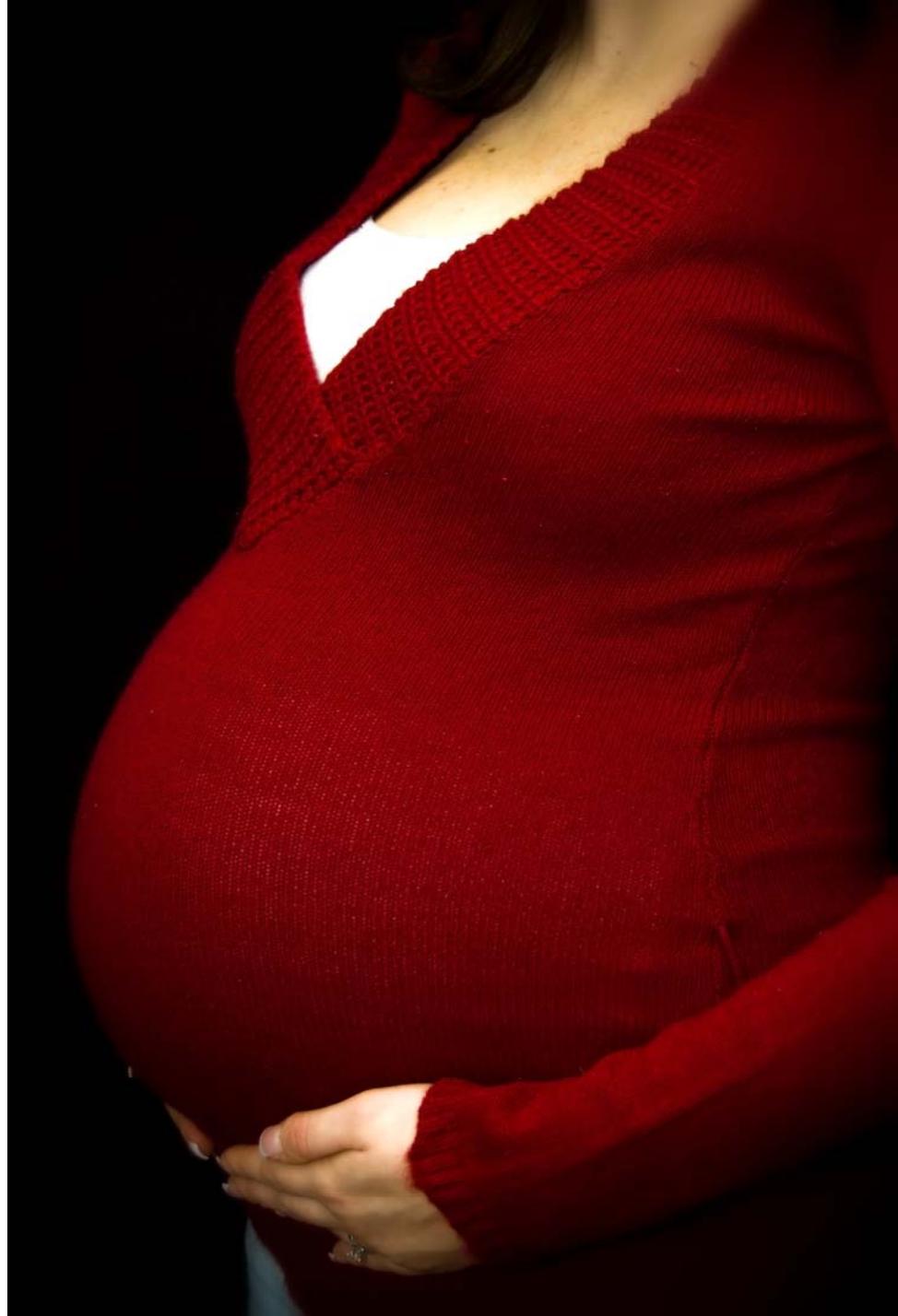
Routinely used for **fetal sexing** in high risk pregnancies

Routinely used for identification of fetal **blood group** - **first clinical application**

Down syndrome (T21) testing: in development in the **UK**

Sequenom: MaterniT21 and **MaterniT21 PLUS** - clinically available in **US**

Direct to consumer (DTC) tests: paternity and fetal sex (**US - global reach**)



Future developments?

Refinement of **T21** testing - **100%** sensitivity and specificity?

Single gene disorders: early stage of development.

Disease **susceptibility** genes, **late onset** diseases, **non medical** traits

Whole genome sequencing?



Practical issues

How might NIPD fit with current clinical pathways?

- Supplementary 'advanced screening' test?
- Replacement screening test?
- Replacement diagnostic test?

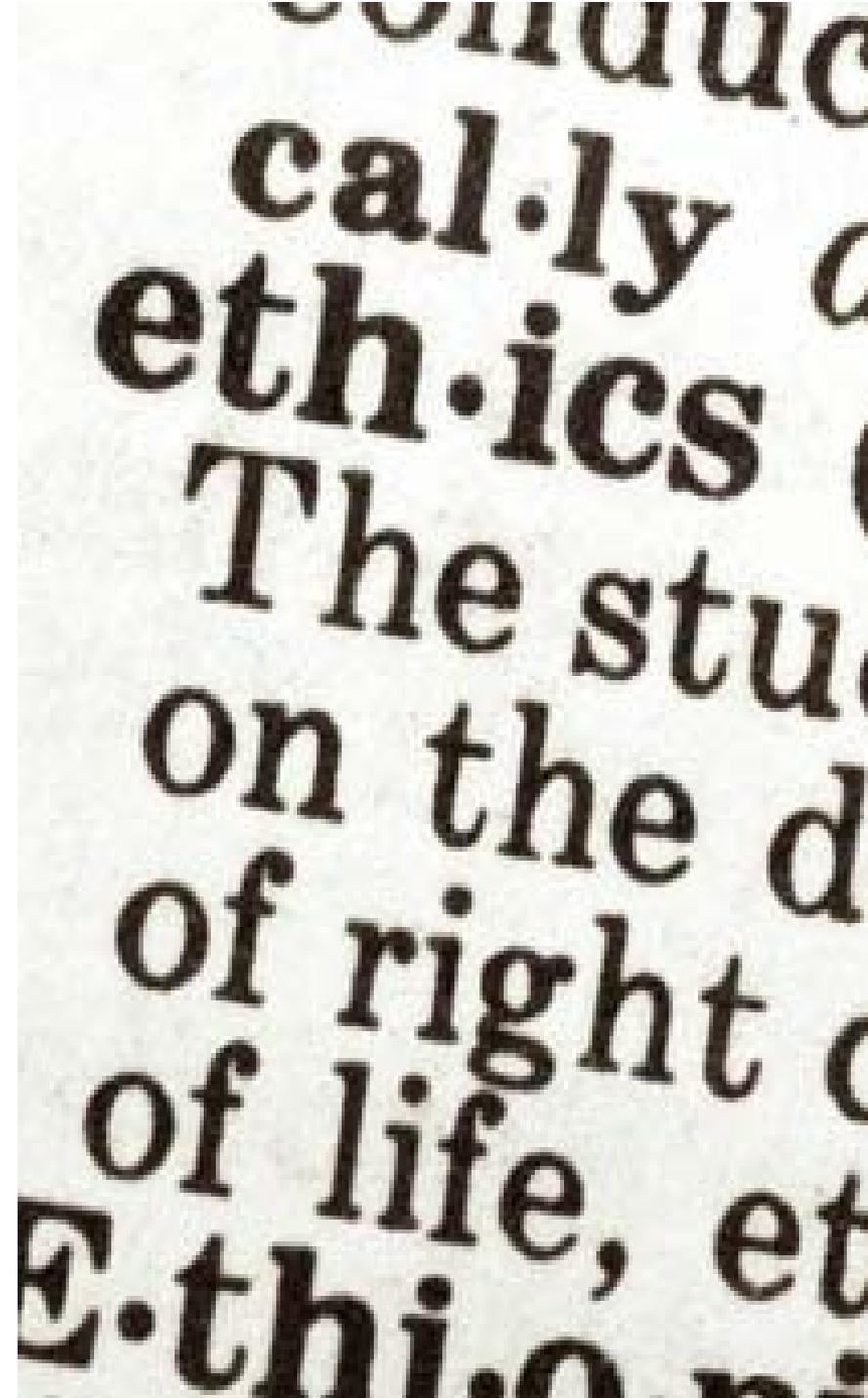
Governance and regulation?

Multiple commercial companies, **global** market, **IP** issues

Private or public funding feasible?

Ethical issues raised

- Informed consent
- Normalisation of testing and termination?
- Specification creep
- Economic and practical burden?
- Disability rights critiques



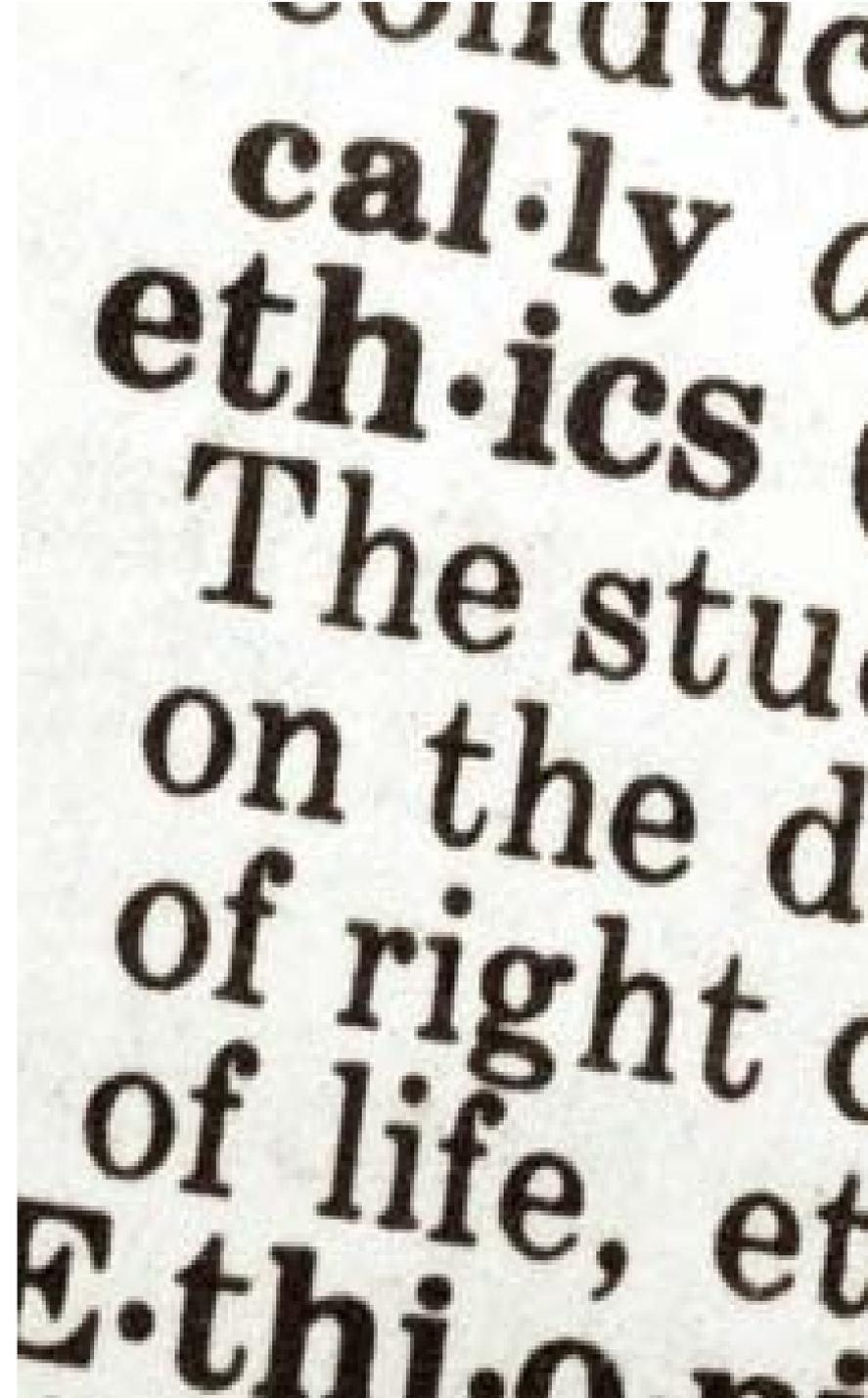
Informed consent

Convenience of NIPD = erosion of informed consent procedures?

“practitioners will view the consent process for prenatal diagnostic testing differently depending on whether it is an invasive or non-invasive test”. Van den Heuvel et al, 2010

Deans and Newson: informed consent for NIPD should mirror procedures for invasive testing

Old debates? Criticism of level of information provided for current screening programmes.



Normalisation of testing and termination

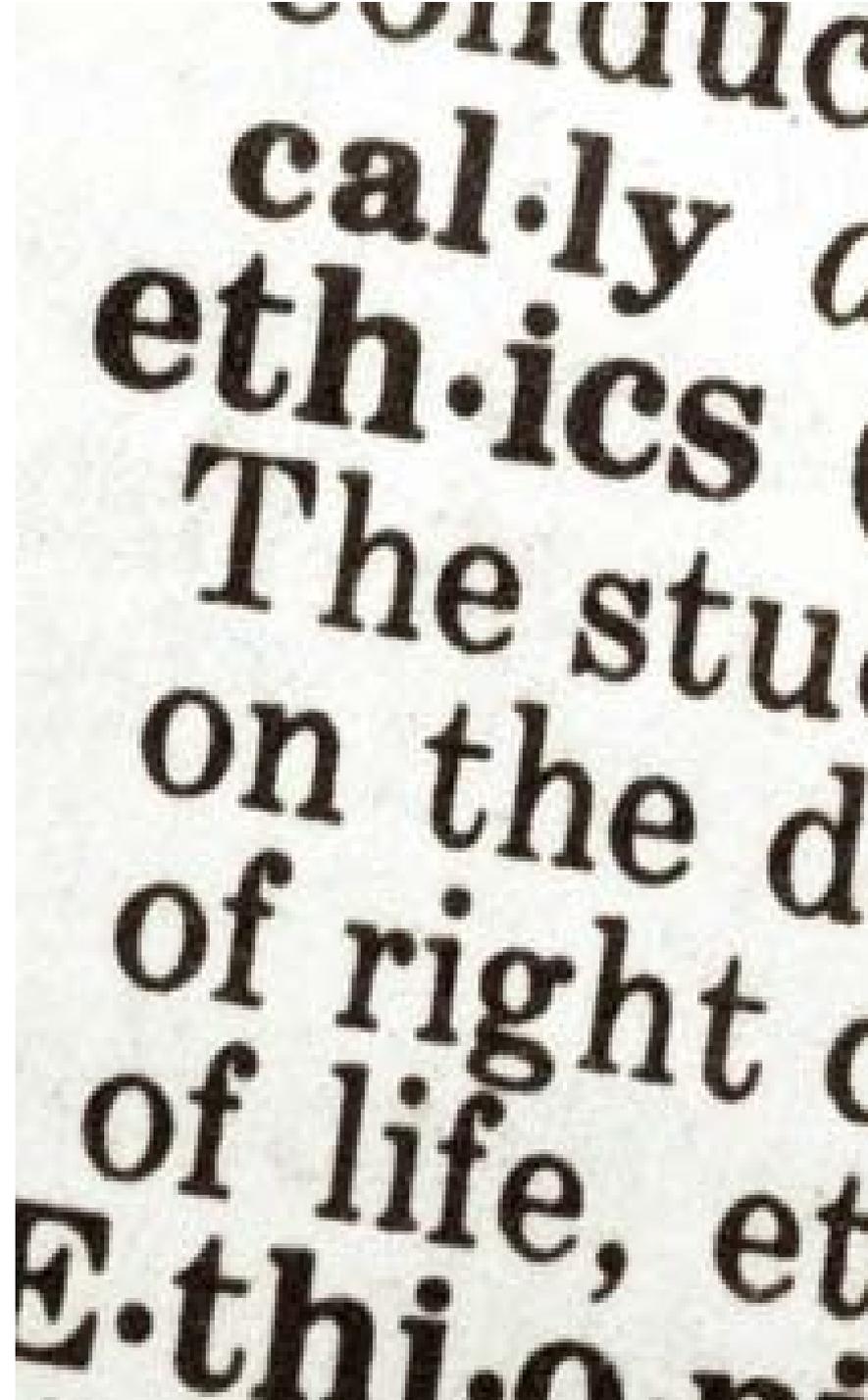
Would NIPD screening be difficult to refuse?

- Ease of testing - noninvasive
- Early stage of pregnancy

Easier to terminate?

- Before pregnancy outwardly obvious
- Before maternal-fetal bonding
- Testing produces no image of the fetus

Psychological and emotional weight of termination underestimated here?



Specification creep

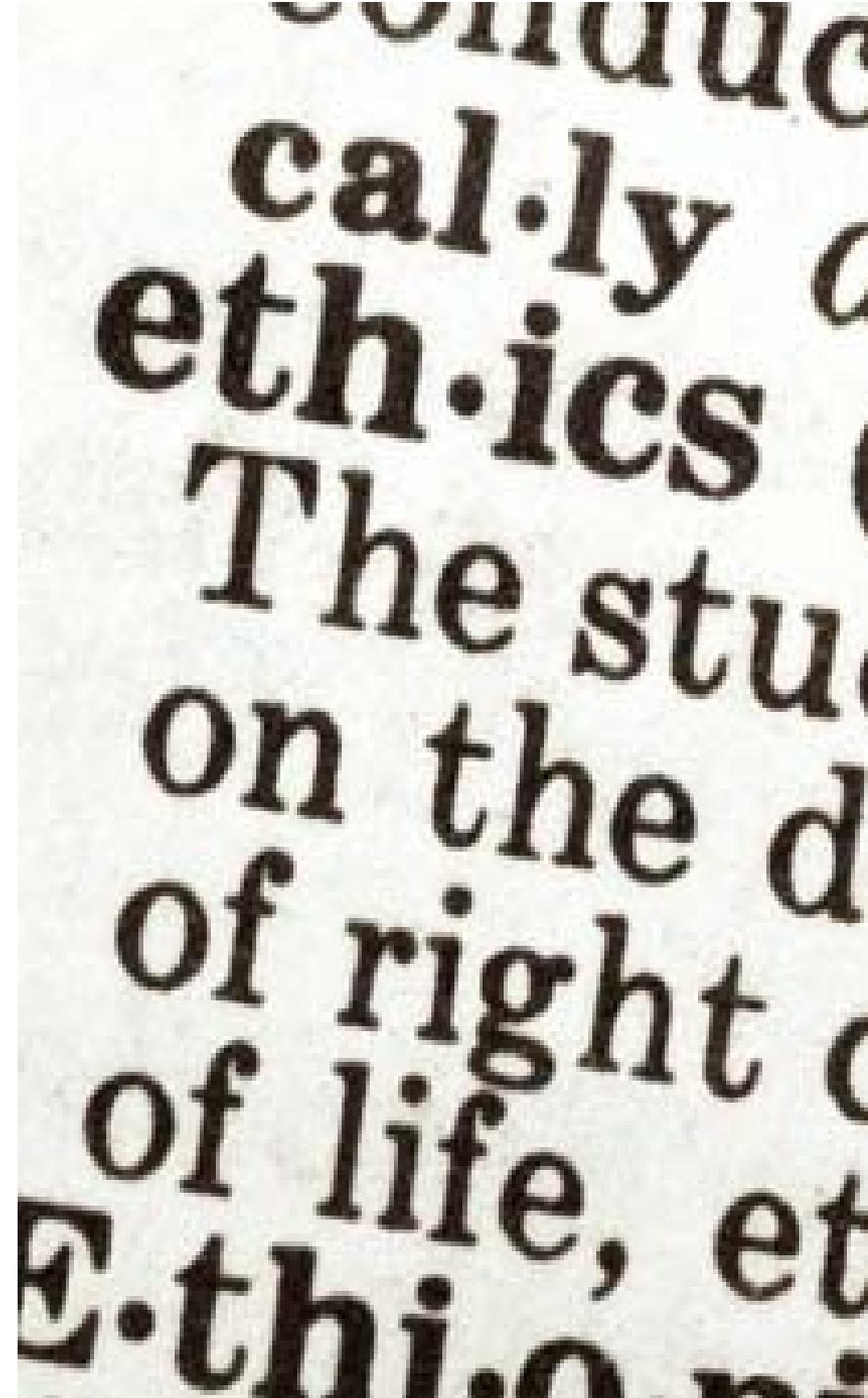
'Slippery-slope' argument. Closely related to discussions of **routinisation**.

NIPD has already moved from blood group testing, to fetal sex, to T21 and now T21 & T13 and T18

Old debate: Late onset disorders? Susceptibility testing? Familiar issues in prenatal genetic counselling literature.

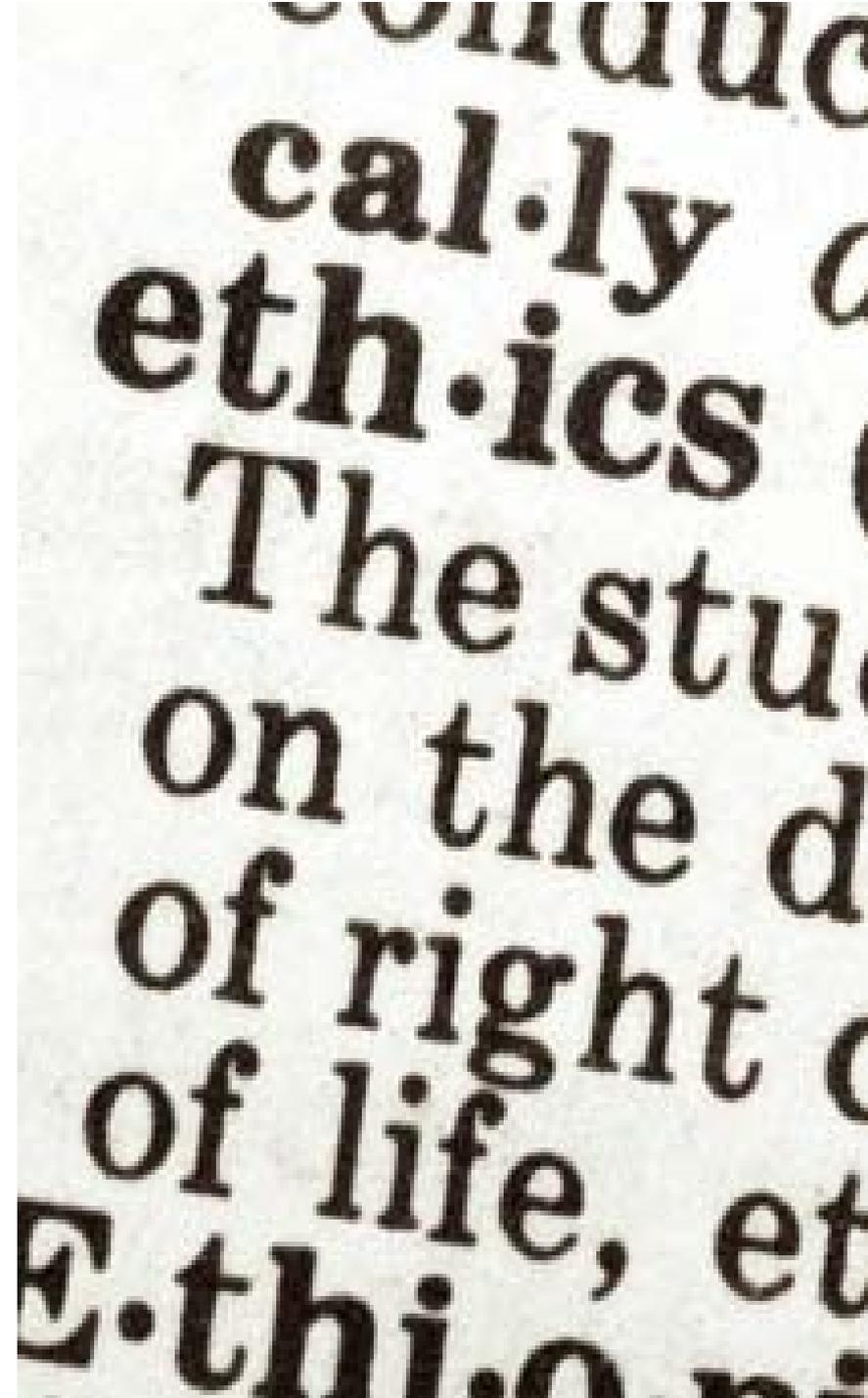
Dystopian futures - widespread **sex selection**, testing for social and non medical traits.

Governance and regulation key issue here. Should testing be regulated? If so, how?



Economic and practical burdens

- Economic cost of population-wide screening programmes
- Population-wide screening: burden of providing pre- and post-test counselling
- Re-training of health professionals



Disability rights critique

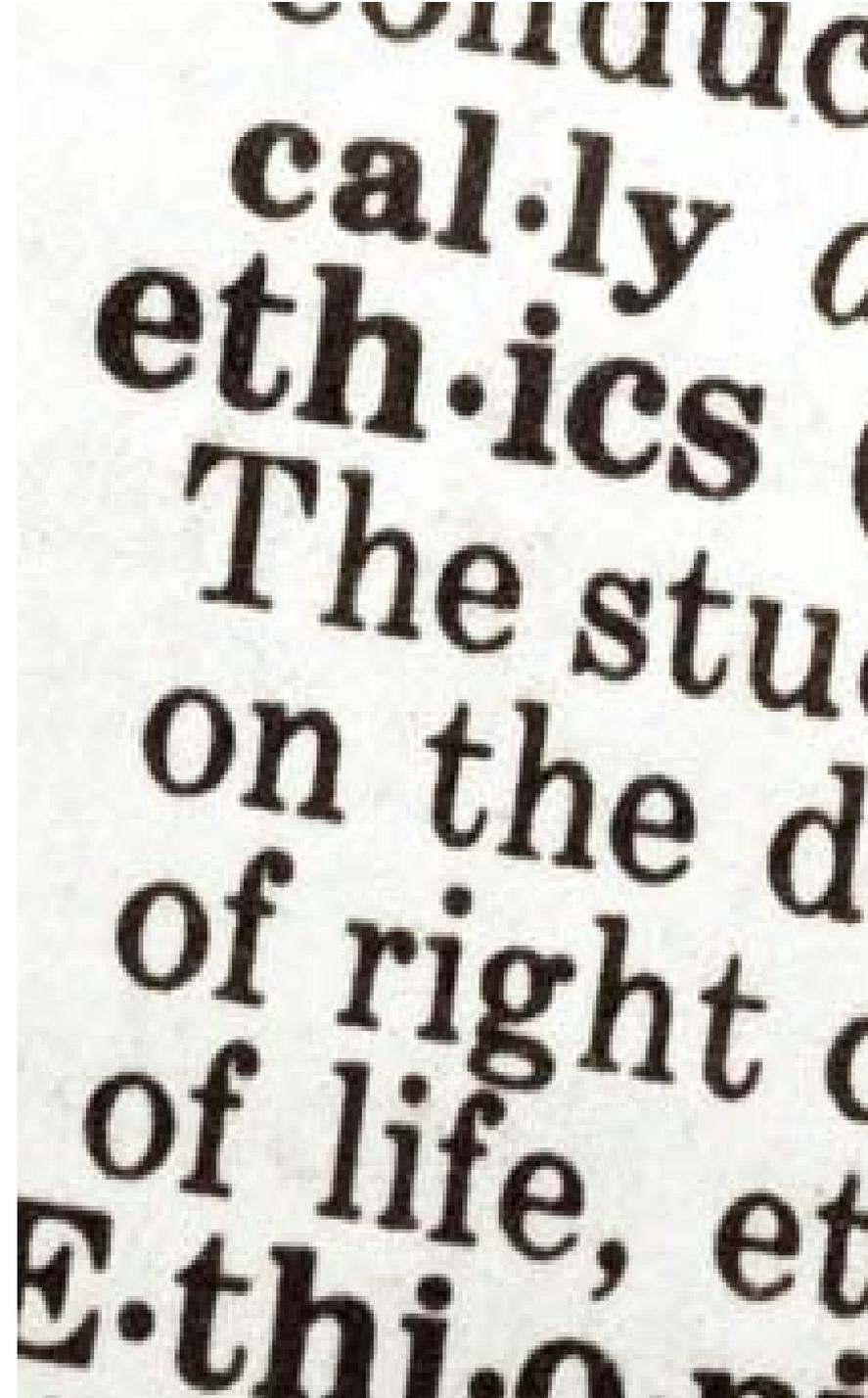
Debate re-ignited by NIPD: “**With the new prenatal testing, will babies with Down Syndrome disappear?**”. Skotko, 2009.

- Respect for those living with condition tested for eroded
- Screening and promotion of the ‘new eugenics’

Highly contentious issue - often **marginalised** within mainstream literature.

Evidence that issue is of **public interest and concern**: Kelly and Farrimond, 2012.

Should assumptions regarding quality of life be reevaluated in light of modern medical and social care?



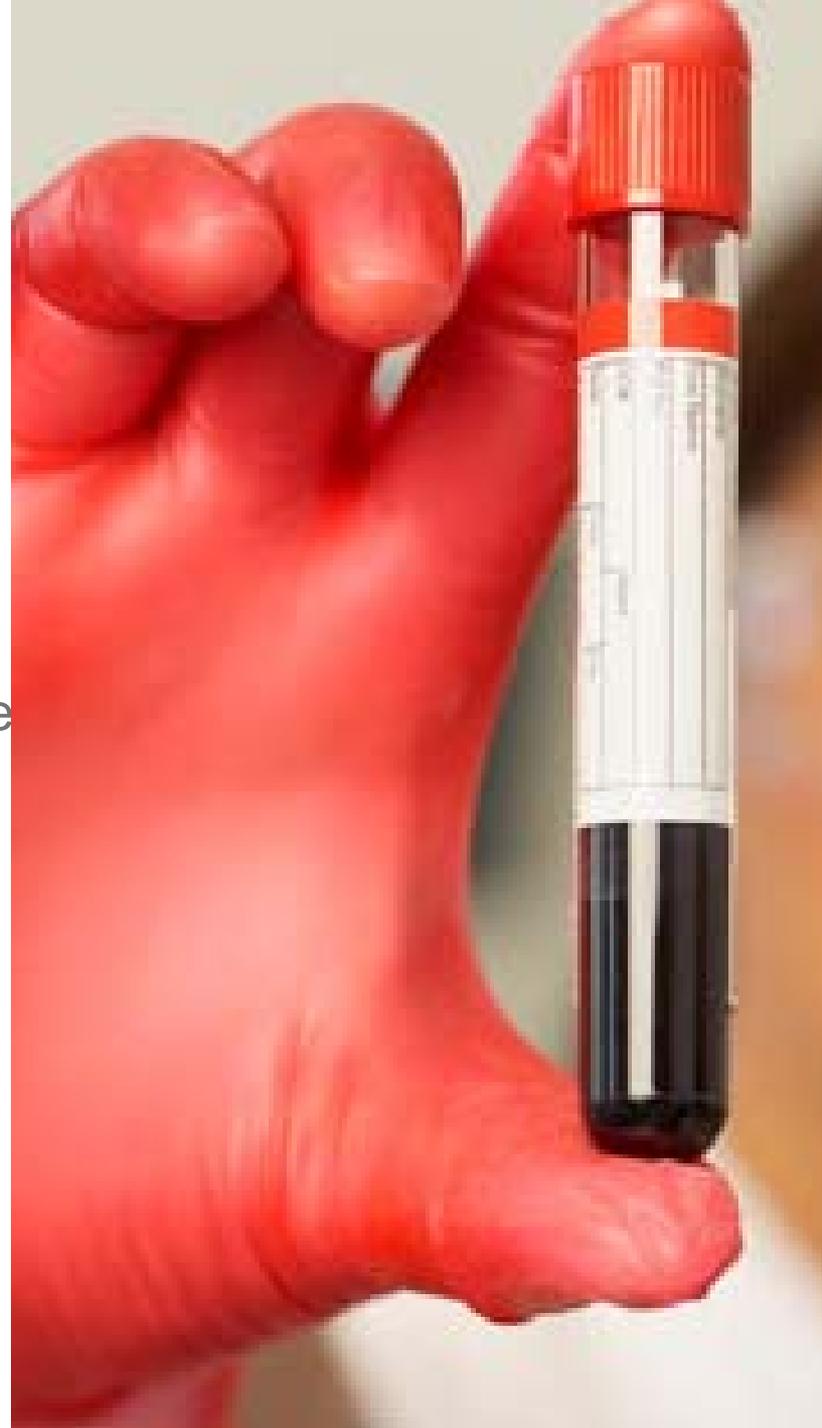
Ethics and experiences of NIPD

Routinisation, normalisation of testing:

“Having that blood test is just nothing, it’s like any of your other visits to hospital when you’re pregnant, it’s just the results that have such a big impact.”

Early testing and maternal-fetal bonding:

“we lost the girl that we were carrying. Obviously I’d started to bond and we had a name and it seemed much more real.”

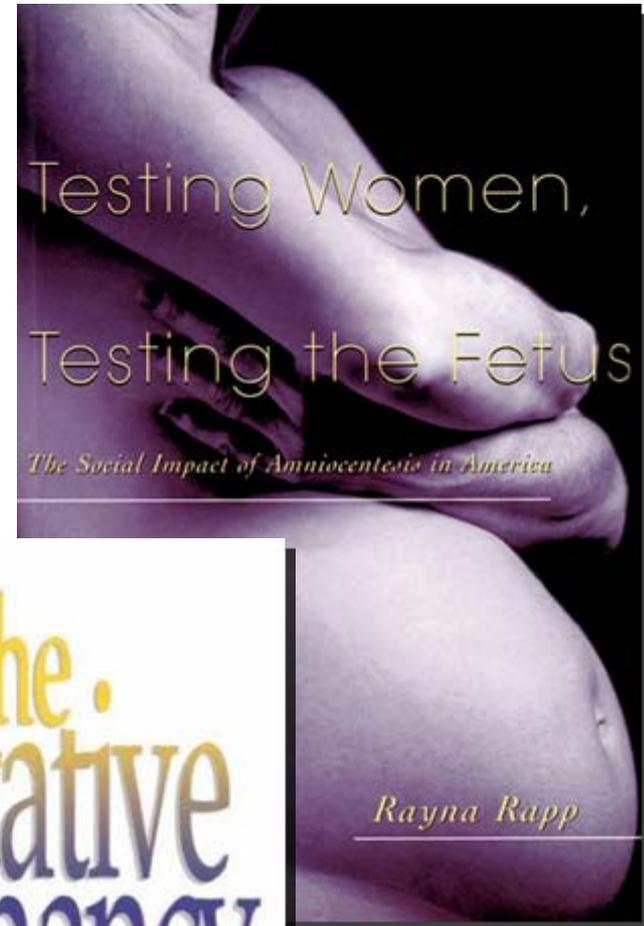


Old debates? Prenatal screening and diagnosis in social science

Valuable relationship between bioethics and social science.

Bioethics: provokes analysis and debate

Social science: contextualises, highlights complexity and recognises value of lived experience



The.
Tentative
Pregnancy
How Amniocentesis
Changes the Experience
of Motherhood
BARBARA KATZ ROTHMAN

Areas for further research

Experiences of NIPD: women, partners, clinicians, genetic counsellors

Public **responses**: interested groups, public at large

Parallel developments in technology - 3D and 4D scanning - ultrasound smartphone - whole genome sequencing - how do technologies **interact**?



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