

# Canaries in the coal mine?

People with disabilities &  
non-invasive prenatal diagnosis



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# Disclosure of speaker interests

<b>(Potential) conflict of interest</b>	<b>None</b>
<b>Potentially relevant company relationships in connection with event</b>	<b>Company names</b>
<ul style="list-style-type: none"><li>• Sponsorship or research funding</li><li>• Fee or other (financial) payment</li><li>• Shareholder</li><li>• Other relationship, i.e.</li></ul>	Member of Nuffield Council on Bioethics

# Overview

- Disability and selective abortion ethics
- UK prenatal screening and diagnosis
- NIPD as screening
- Costs and benefits of NIPD
- Future developments
- Ethical questions
- Nuffield Council on Bioethics work

# Is selective abortion different from family planning abortion?

- Difference between “not wanting to be pregnant” vs “not wanting to be pregnant with this fetus”?
  - Choosing on the basis of characteristics of the fetus
  - Discriminatory towards disabled people
  - Expressivist argument
- Early termination vs late termination: moral and psychological weight

# My position on PND (Shakespeare 1998)

- Support a woman's right to choose (gradualist)
- Need to support women/couples to have test or refuse test; to continue pregnancy or to terminate
- Need to support women who opt for termination
- Need to provide full and balanced information about all aspects of living with disability
- Need to welcome babies with disability, support families, remove barriers

# PND: *Ethical versus Eugenic*

- Individuals and families
  - Voluntary
  - Informed consent
  - Goal: reproductive autonomy
  - Population policy
  - Coercive
  - Goal: elimination of disability
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- But: historical eugenics also involved a voluntary component.
  - 'Laissez faire eugenics' (Kitcher); eugenics as 'emergent property'
  - Empirical evidence challenges extent to which informed consent applies: routinisation, lack of balanced information, implicit directiveness
  - Ambiguity as to whether reproductive autonomy or prevention of affected babies is key motivation: cost/benefit dimension

# Current UK screening

- Serum screening (all pregnant women offered)
- Ultrasound (all pregnant women offered)
- Combination of serum and nuchal translucency measurement gives raised risk of having a pregnancy affected by Down syndrome
- Diagnostic testing offered at 18 weeks, followed by offer of termination of pregnancy
- ~90% of affected pregnancies terminated.

# Current UK diagnostic services

- Where family history of e.g. HD or haemophilia or recessive condition (CF, MD), or raised risk for ethnic group (Thalassaemia, Tay-Sachs etc)
- Individualised counselling
- CVS or amnio followed by termination of affected pregnancies
- Pre-implantation genetic diagnosis
- Not offered to wider population



# Non-invasive prenatal diagnosis

- Cell-free DNA – 10% of cffDNA in maternal plasma of fetal origin – detectable at 7-10 weeks gestation
- Began to be offered commercially from 2011
- Diagnostic for sex, rhesus, and some monogenic conditions. WGS technically possible.
- Screening test for chromosome anomalies: false positive 0.09% and detection rate 99.2% for trisomy 21, a bit worse for t18 and t13.
- May also reveal cancer diagnoses in the women

# Benefits of NIPD: PND “Holy Grail”

- Non-invasive: reduces risks of miscarriage (~1%)
- Reliable, not painful, easy to perform
- Early – 10<sup>th</sup> week of pregnancy: reassurance
- Reduces number of women who need to have a diagnostic amniocentesis, with risk of fetal loss (2015 Pegasus study in Canada predicts reduction in amnio from 10,000>300, fetal loss 70>1)
- Also reduces health care costs
- Earlier abortion



“Hundreds of babies could be saved after Down’s Syndrome blood test is approved for NHS”

15 January 2016



“Blood test for Down's set to be offered on the NHS: Super-safe and 99% accurate exam would replace invasive procedure which can trigger a miscarriage”

15 January 2016



“Safer Down's test backed for NHS use: Pregnant women in the UK should soon get a safer and more accurate test for Down's syndrome on the NHS, to reduce the risk of miscarriage.”

15 January 2016

# NHS response

- NHS National Screening Committee recommends NIPT as a second stage screen only for women of risk of 1:150 or higher
  - Would detect 102 more DS pregnancies
  - Lead to 4,870 fewer invasive tests
  - 25 fewer miscarriages
  - £337,000 p.a. lower cost to NHS (test cost £250)
    - (England and Wales figures)
- Offering it to everyone would detect more DS pregnancies, but not reduce miscarriages and would cost up to £8m more. (Chitty et al 2015)

# Does this change the ethical assessment?

- Non-invasive - so lower threshold for adoption; maybe also less counselling provided. No need to discuss fetal loss.
- But also confusion over new test.
- Earlier in pregnancy – hence earlier abortion if positive (Dar et al 2014 found 6.2% proceeded to termination without diagnostic confirmation)
- But also provides early information for those who would continue
- Further routinisation of PND – may reduce possibility of saying no: may stigmatise those who continue pregnancies, people with disabilities?
- Inequality of access to NIPT

# Future developments

- NIPT for sex selection (as early as 7 wks)
- Wider extension of NIPT PND: late onset conditions; minor conditions; carriers; non-medical conditions
- NIPT plus WGS
- If NIPT improves accuracy, becomes diagnostic: early medical abortions on wider grounds at 10 wks
- Direct to Consumer Testing – without counselling
- Current cost ranges from \$350-\$3,900 globally
- Inequality of access to NIPT – reproductive tourism, health inequities

# Does this change the ethical assessment?

- NIPT makes prenatal diagnosis and selective termination quicker, easier and less morally troubling for Down syndrome
- NIPT+WGS lowers the bar for prenatal diagnosis and selective termination for other conditions: trivial, non-clinical
- Do we think again about morality of choosing on the basis of characteristics of the fetus?
- Or do we accept the implications of consistent disinterest about morality of early termination decisions?

# Nuffield Council on Bioethics project on NIPT

- Working group (not working party with 2 year timescale and book-length report)
- Louise Bryant (psychologist), Tara Clancy (genetic counsellor), Zuzana Deans (philosopher), Angus Clarke (clinical geneticist), Shaun Pattinson (lawyer)
- Survey and consultation (public and expert) on website (respond by 25 July)
- Meetings with expert groups
- Publication of report in February 2017



# References

- Chitty L et al, RAPID non-invasive prenatal testing (NIPT) evaluation study: executive summary, 2015.
- PEGASUS (personalized genomics for prenatal aneuploidy screening using maternal blood) study <http://pegasus-pegase.ca>
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- Taylor-Phillips S et al, Systematic review and cost-consequent assessment of cell-free DNA testing for T21, T18 and T13 in the UK – final report, 2015.