GNBS Ethical Considerations

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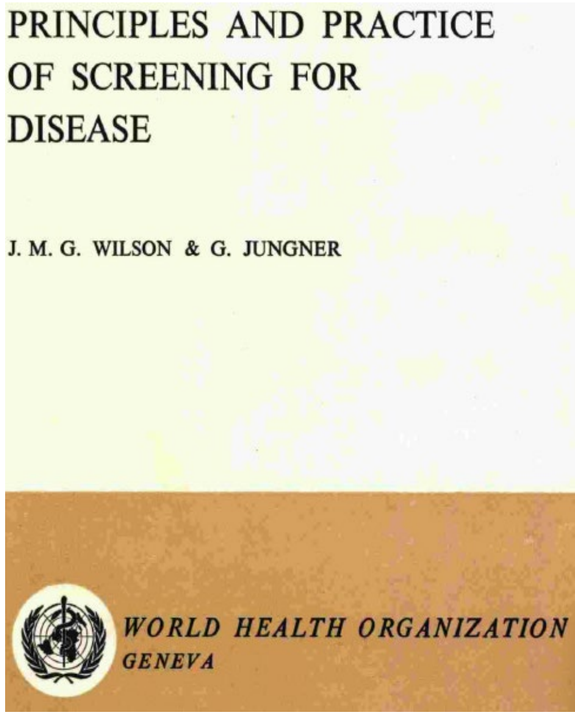
Skepticism toward GNBS

Ethical concerns include:

- Informed Consent – how to help people make informed choices about gNBS
- Data Privacy – how to ensure data generated is used carefully
- Inaccurate Results – false positives/false negatives
- Uncertainty of Results – VUS could cause additional stress and uncertainty
- Cost-Effectiveness – potential impacts could overwhelm the health system
- Opportunity Costs – may replace more effective screening methods

Is GNBS ethically distinctive?

- Different in degree, but not in kind, compared to other screening tests
- Concerns will be context-sensitive—population, disease, resources
- Justifies current use of GNBS in a research setting.
- Ethical approach is the same



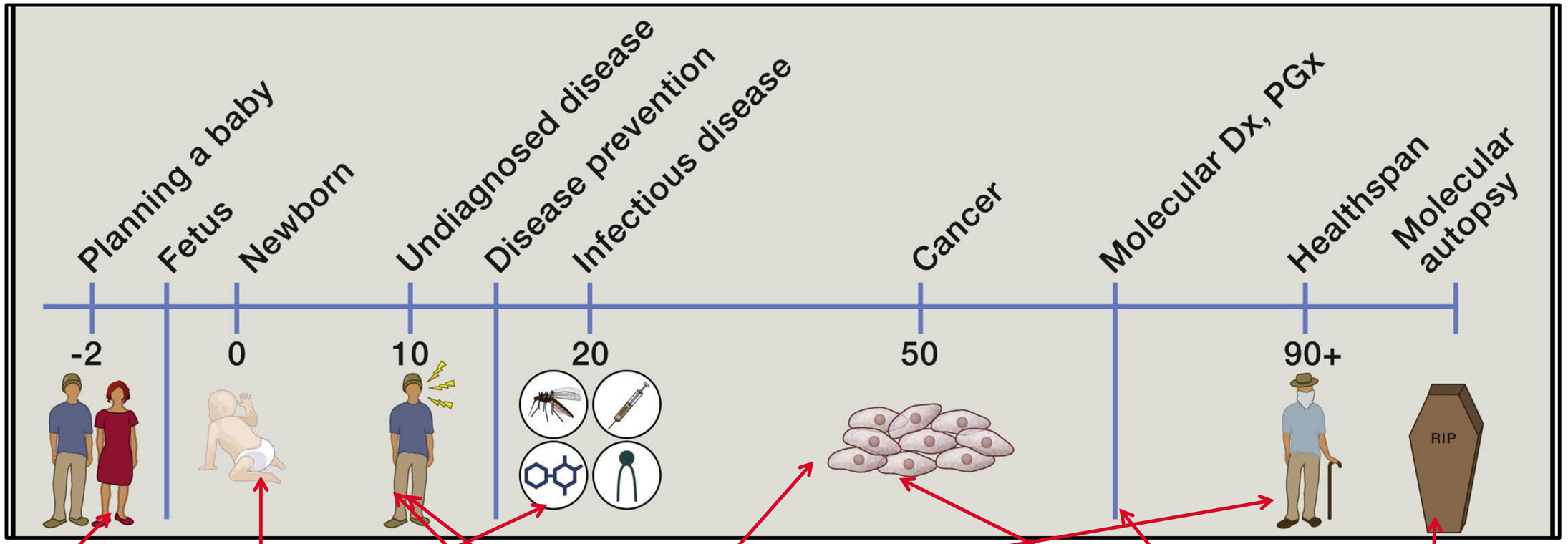
"The benefit gained by individuals from the screening programme should outweigh any harms, for example from overdiagnosis, overtreatment, false positives, false reassurance, uncertain findings and complications."
(UK population screening guidelines)

UK National Screening Committee

The opportunity cost of the screening programme (including testing, diagnosis and treatment, administration, training and quality assurance) should be economically balanced in relation to expenditure on medical care as a whole (value for money).

Sequential interrogation

Genomic sequencing at birth
- re-interrogate the data as new clinical questions arise



reproductive carrier testing

second tier to newborn screening

cancer patients – hereditary cancer prediction; precision therapies

prevention of common (complex, polygenic) disease

predisposition testing for treatable monogenic disorders

childhood or adult onset disorders suspected of being genetic (incl. ultra-rapid genomic testing)

pharmacogenomic testing

molecular autopsy

from Topol E, Cell 2014

Genomic data as a health resource

Reframes the Relevant Ethical Considerations

- Data generation vs data use
- Each data use case should be considered separately
- Maximize the clinical/research potential of genomics
- Raises questions about the economics of storing versus sequence on demands.

Extended essay

Storing paediatric genomic data for sequential interrogation across the lifespan

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Journal of Medical Ethics

The Human Rights Argument for Sequential Interrogation

International Covenant on Economic Social and Cultural Rights

Article 12
The States Parties to the present Covenant recognize the right of everyone to the enjoyment of the highest attainable standard of physical and mental health.

Article 15
The States Parties :
(b) To enjoy the benefits of scientific progress and its applications

Article 6
States Parties shall ensure to the maximum extent possible the survival and development of the child

UDHR
All people should enjoy human rights "without distinction of any kind, in particular as to race, colour or national origin"

Genomic Privacy Project

Lineage Consortium Targeted Research Project
(with Ainsley Newson, Margaret Otlowski, Yingyi Luo, Clara Gaff, Zornitza Stark,, Tom Conway, Gabby Chandler)

Privacy as a Key Consideration in Genomic Data Governance

- Sensitive and personal?
- Genetic exceptionalism?

Project Focus Areas:

- When do we lose genetic privacy (forms of genomic data)?
- When is genomic data is a privacy risk?
- How might new technologies help protect privacy?

AUSTRALIA

Privacy Act 1988

No. 119, 1988

Trust and Genomics Project

Lineage Research Project
(with Ainsley Newson, Lisa Dive)

Trust as Essential in Genomic Data Governance Frameworks

- Sometimes viewed as a prerequisite for genomics research
- Sometimes seen as a target or aim

LINEAGE Study

Project Focus Areas:

- Different ways to conceptualize trust
- Look at ‘degrees’ of trust
- How different forms of trust are important
- Exploring various methods for fostering trust

Thanks