

# Ethical Issues in Genetics Testing/Research

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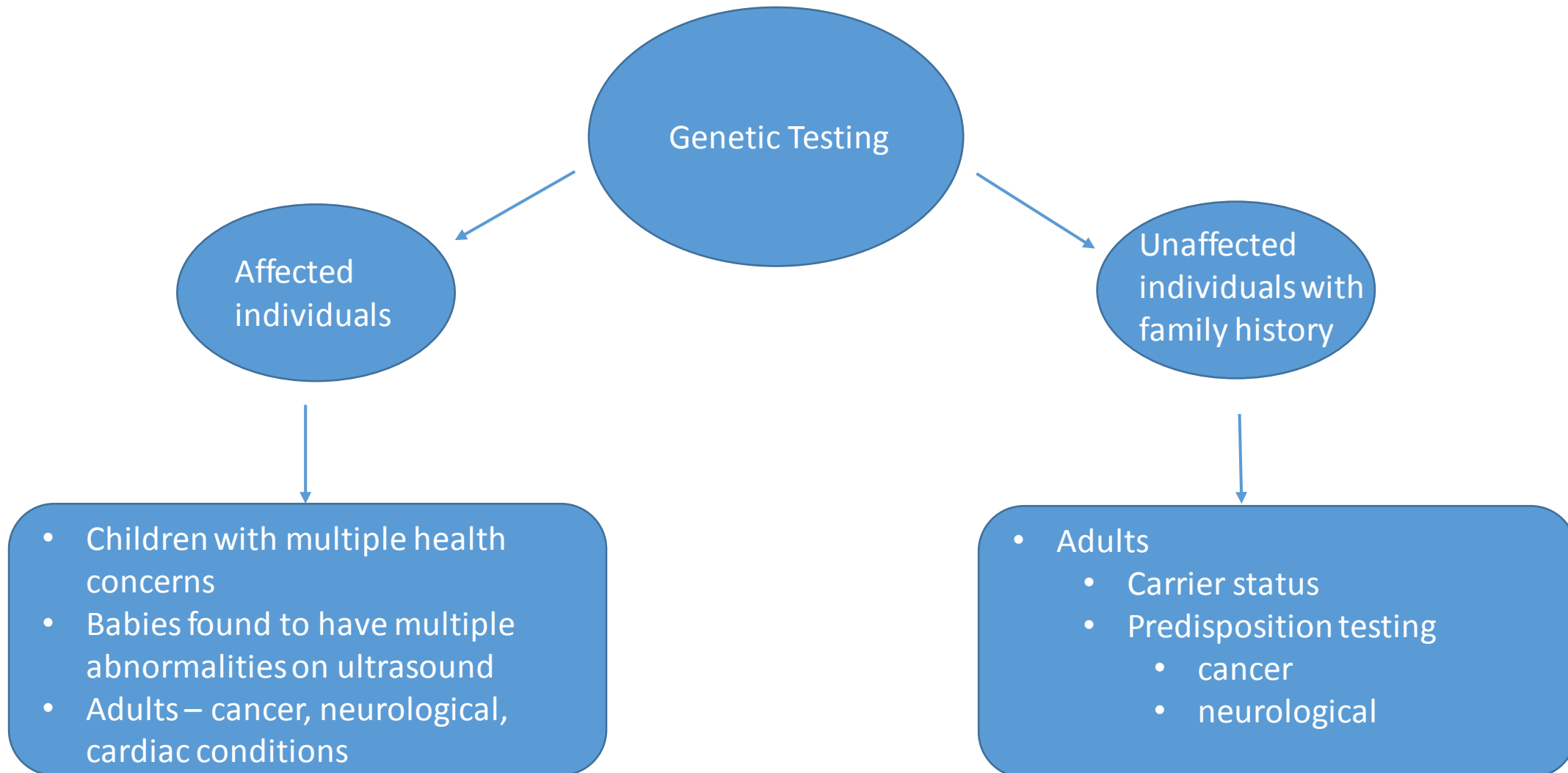
# Medical Ethics principles

- Principle of respect for autonomy
- Principle of nonmaleficence
- Principle of beneficence
- Principle of justice

# Ethical issues related to Genetic Testing

- Who is being tested?
  - Affected with a genetic/possibly genetic condition
  - Unaffected but at increased risk
- How are they being tested?
  - One “gene” approach
  - Multiple/All genes approach
- Consent and Results – who and how?

# Who is/was being tested?



# Ethical issues associated with testing one “gene” at a time for this group

- Privacy and Confidentiality – shared nature of genetic information
- Informed consent and results disclosure
  - Affected
    - Diagnosis of a genetic condition
    - Inherited? Parental Guilt – especially when came from one parent
    - Recurrence risk – parents, child, extended family
    - Prognosis? Associated with other complications?

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  - Affected
  - Unaffected
    - **Carrier status – impact on family planning**

# Ethical issues associated with testing one “gene” at a time for this group

- Privacy and Confidentiality
- Informed consent and results disclosure
  - Affected
  - Unaffected
    - Carrier status – impact on family planning
    - **Predisposition to hereditary cancer**
      - Types of cancer and risks of developing each
      - Screening/Prevention
      - Risk to children/siblings
      - Impact on family relationships – preselection, survivor guilt
      - Risk of Genetic Discrimination

# What is Genetic discrimination?

- Differential treatment of **asymptomatic** individuals or their relatives on the basis of actual or presumed genetic differences
- It can occur in social situations, insurance (health and life) and employment settings.
- Studies have demonstrated
  - Many experienced GD, small minority sought legal action
  - Fear of GD deters - genetic services and research.
  - Physicians also concerned about GD for their patients – affecting referrals
  - Use of genetic information in underwriting



# What protects people from Genetic Discrimination?

- UNESCO (1997) – Universal declaration on Human Genome and Human Rights
- European Unions Charter of Fundamental Rights (2000)
- UK – Human Genetics Commission – Inside Information (2002)
  - Moratorium Ass British Insurers – can obtain up to 500K insurance without releasing genetic test results
- US – Genetic Information Nondiscrimination Act (GINA)(2008)
  - Healthcare insurance and employment. Not life insurance .

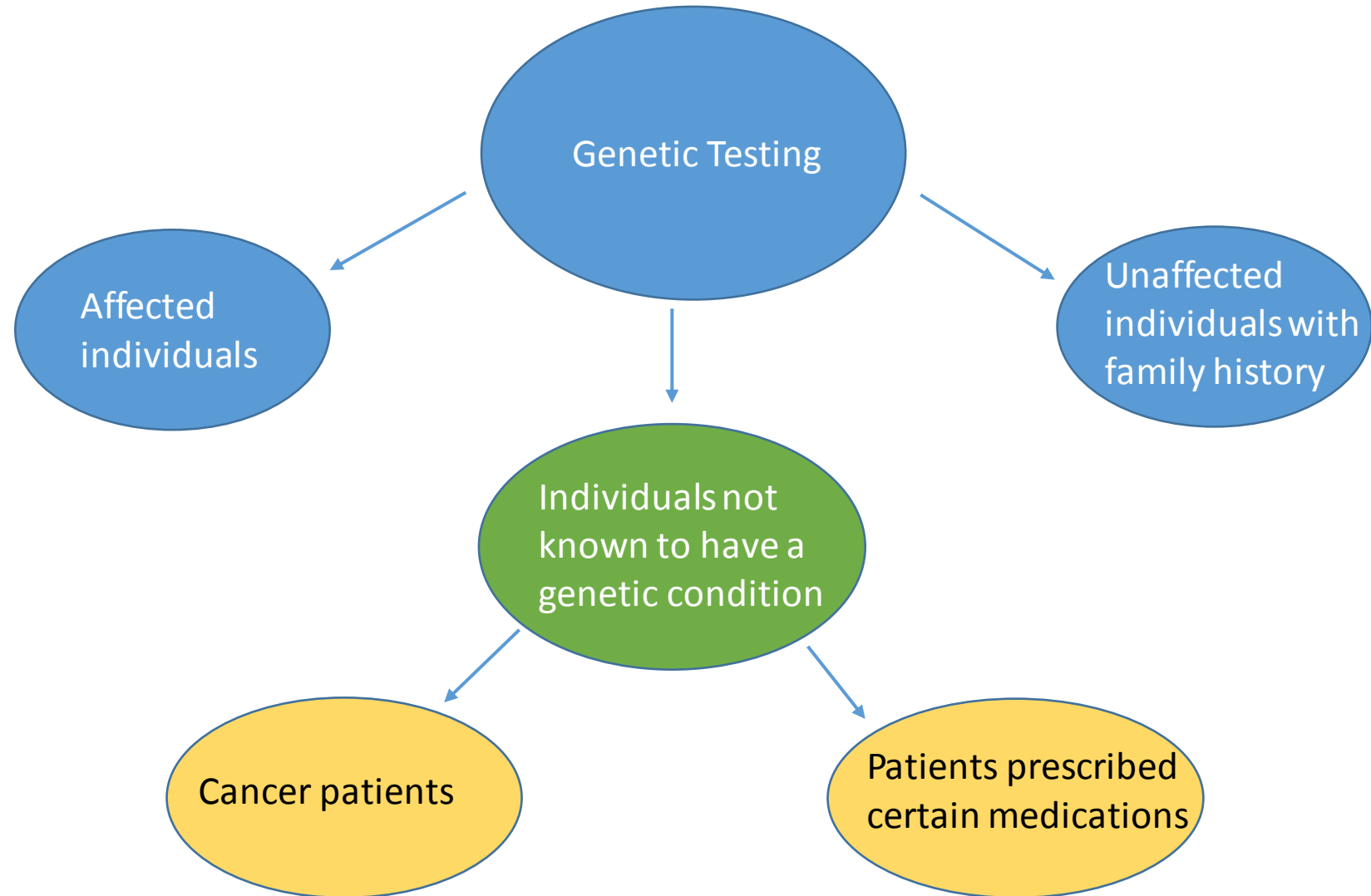
# What protects Australians?

- Federal *Disability Discrimination Act* (1992) - employment
  - 2008 amendment to specifically mention genetic predisposition
- Partial Moratorium Australia and New Zealand – insurers can't request applicant undergo testing but may request results
- QLD state *Anti-Discrimination Act* (1991) has not been modified
- Federal law coverage unclear, no mention of sequencing 100's/1000's

Weisbrot D New Genet Soc 2003: Otlowski et al *Comp Labor Law and Policy Journal* 2010:

Otlowski et al *Ann Rev Genomics Hum Genet* 2012: Keogh and Otlowski 2013: Joly et al *BMC Medicine* 2013:

# Who is going to be tested?



# How are people going to be tested?

- Looking at the code at 1000's of “points” – genotyping – pharmacogenetic testing
- “Panels” of genes – 100's or 1000's
- Code of all the genome – whole genome sequencing – 21,000 genes

# Ethical Considerations Sequencing 100's or 1000's genes

- Informed consent
- **Types of results**
  - Relevant to diagnostic question
  - Variants of unknown clinical significance (**variants uncertain significance**)
  - Clinically or socially relevant for individual/family but not relevant to diagnostic question (**incidental findings**)

# Conclusion

- Genetic Testing always had ethical considerations
  - Shared nature of genetic information
  - Informed consent and optimal result disclosure
  - Genetic Discrimination
- New Technologies
  - Risk of incidental findings
  - Risk of variants of uncertain significance