The state of play

Genetic testing in Australia

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Royal College of Pathologists of Australasia

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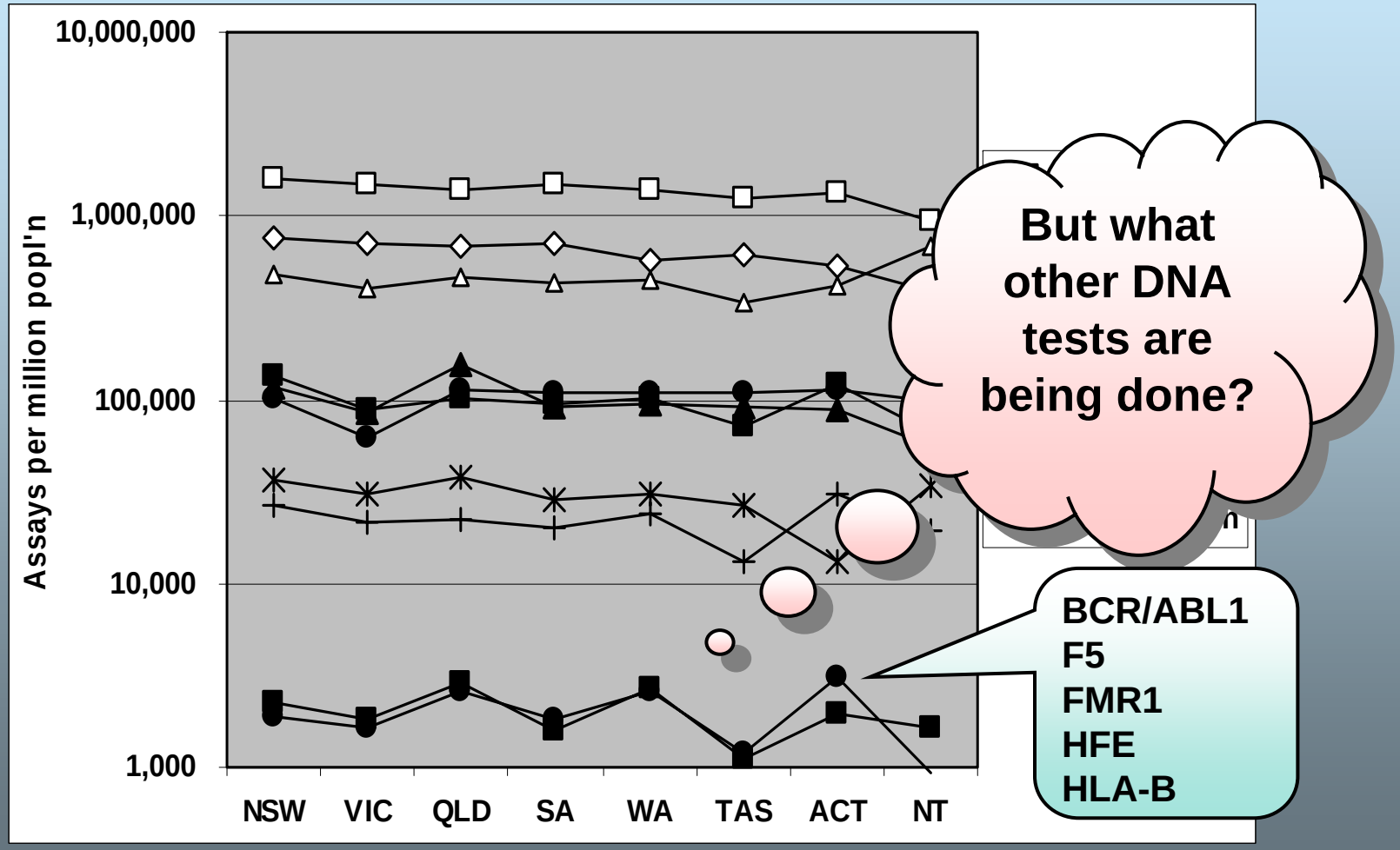
Demand for MBS-funded tests



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The why and how

- Survey of genetic testing offered by Australian labs during 2006.
- Counted number of types of test ("genes") offered.
- Counted volume of assays for each gene.
- Collected non-MBS funded testing from labs.
- Collected MBS-funded data from Medicare website.
- 56 labs identified which provided non-Medicare molecular genetic tests. 93% provided data.
  - 60% public sector
  - 20% private sector
  - 20% academic

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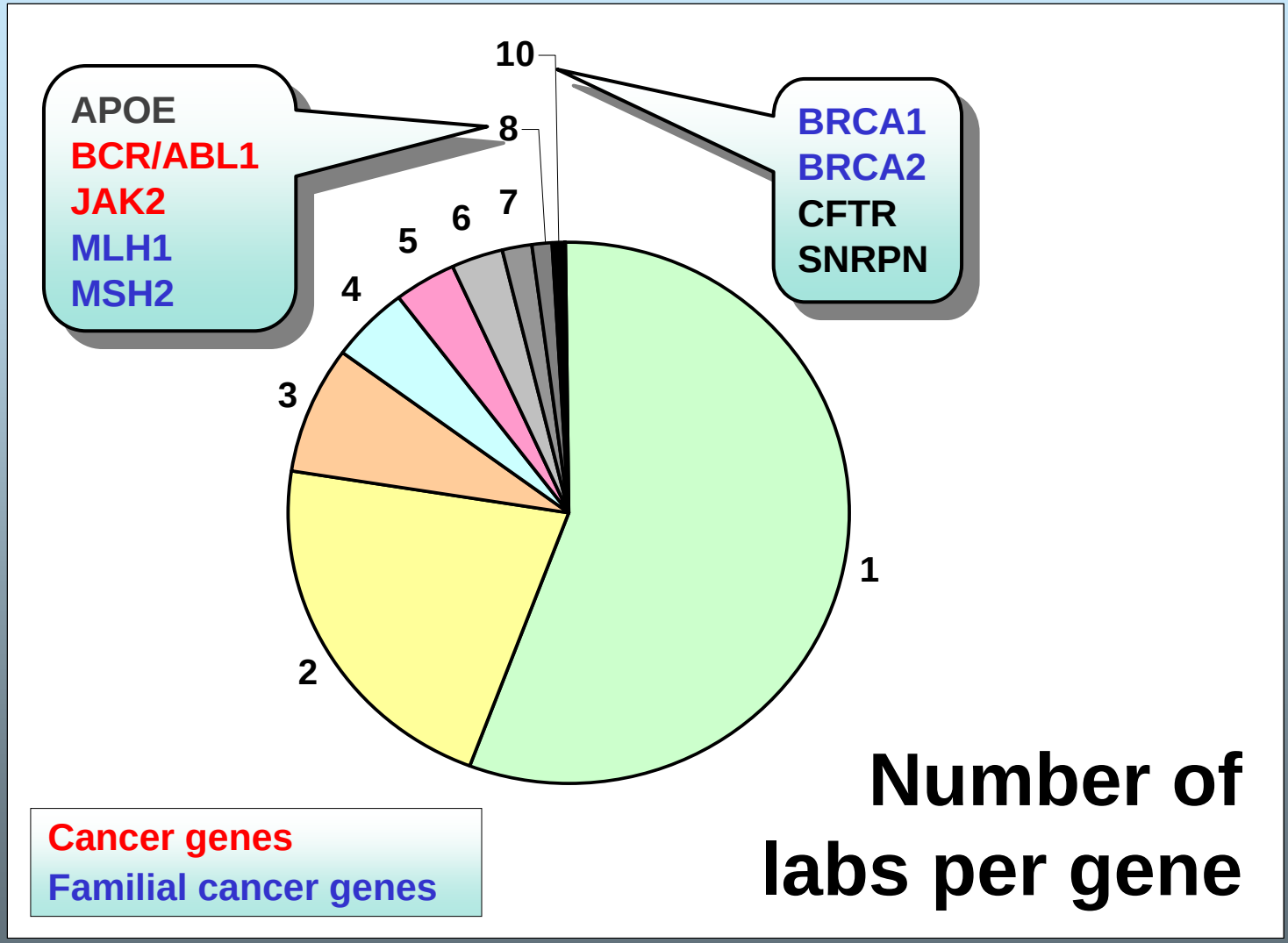
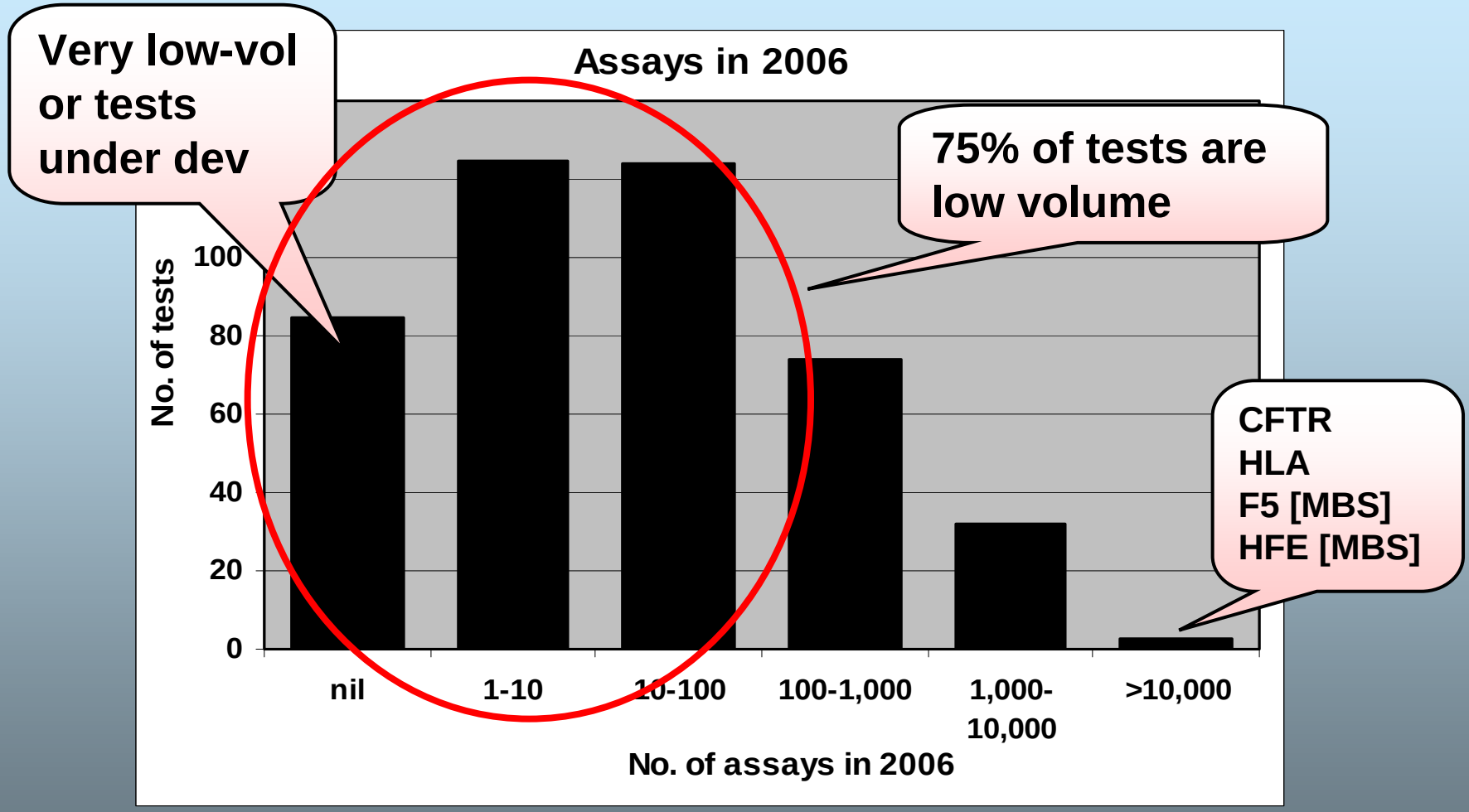
437 genes i.e. types of tests



No. of assays per gene



Medicare	41,497	(26%)
Non-Medicare	119,354	(74%)

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Reason for test

Patient Group	*Assays (%)*	*Genes (>1,000 each)*
Screening	40%	CFTR HLA HEXA
Diagnostic	28%	F5[MBS] CFTR HBA HBB HFE SERPIN1A sub-tel del'n
Somatic	8%	BCL2 BCR/ABL1 IGH@ TRB@ TRG@
Family	5%	CFTR F5
Pharmacogenetic	<1%	
Supplementary	<1%	
Unknown	18%	HFE[MBS] FMR1[MBS]
Total	100%	

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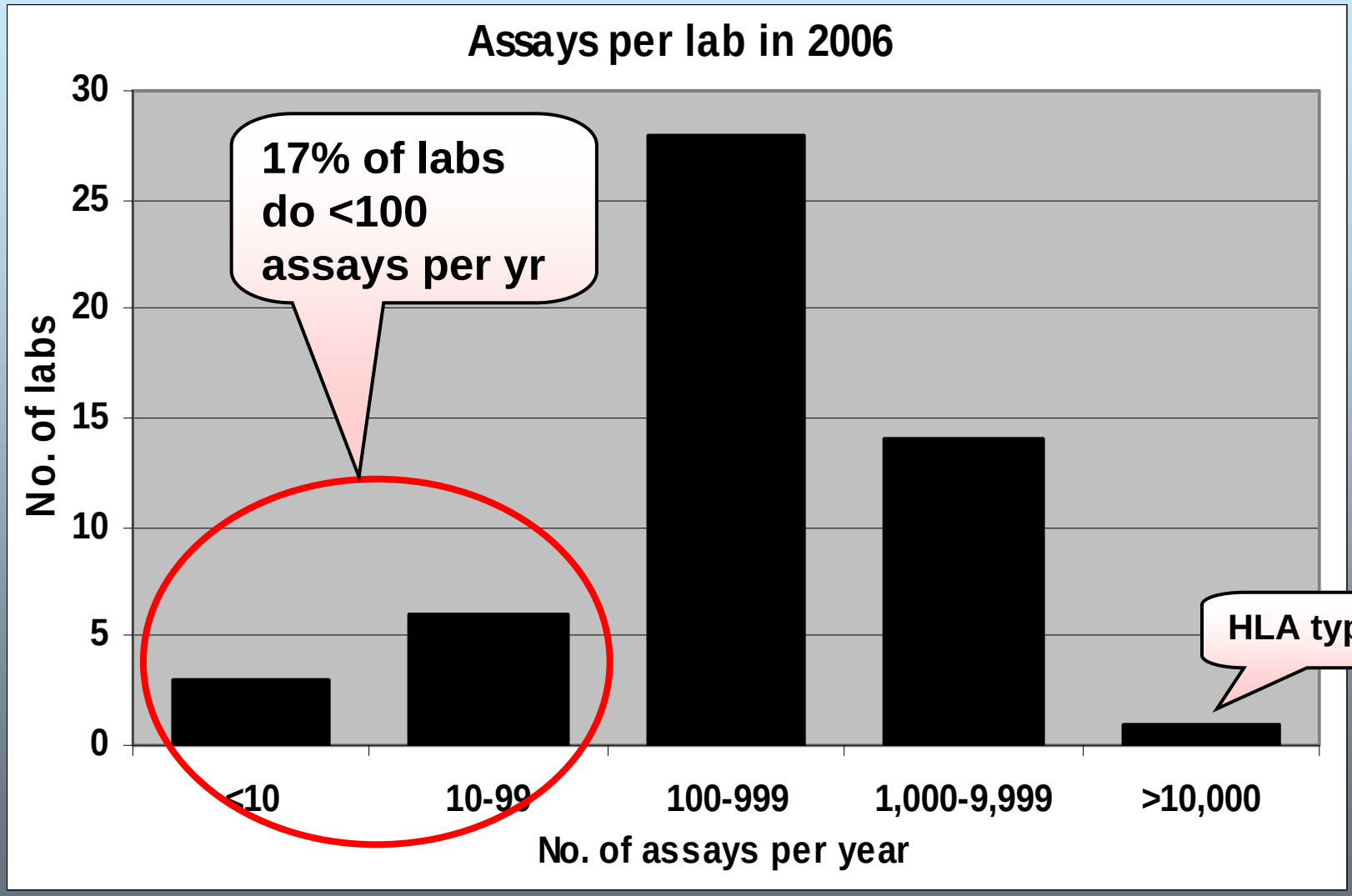
Total assay volume per lab



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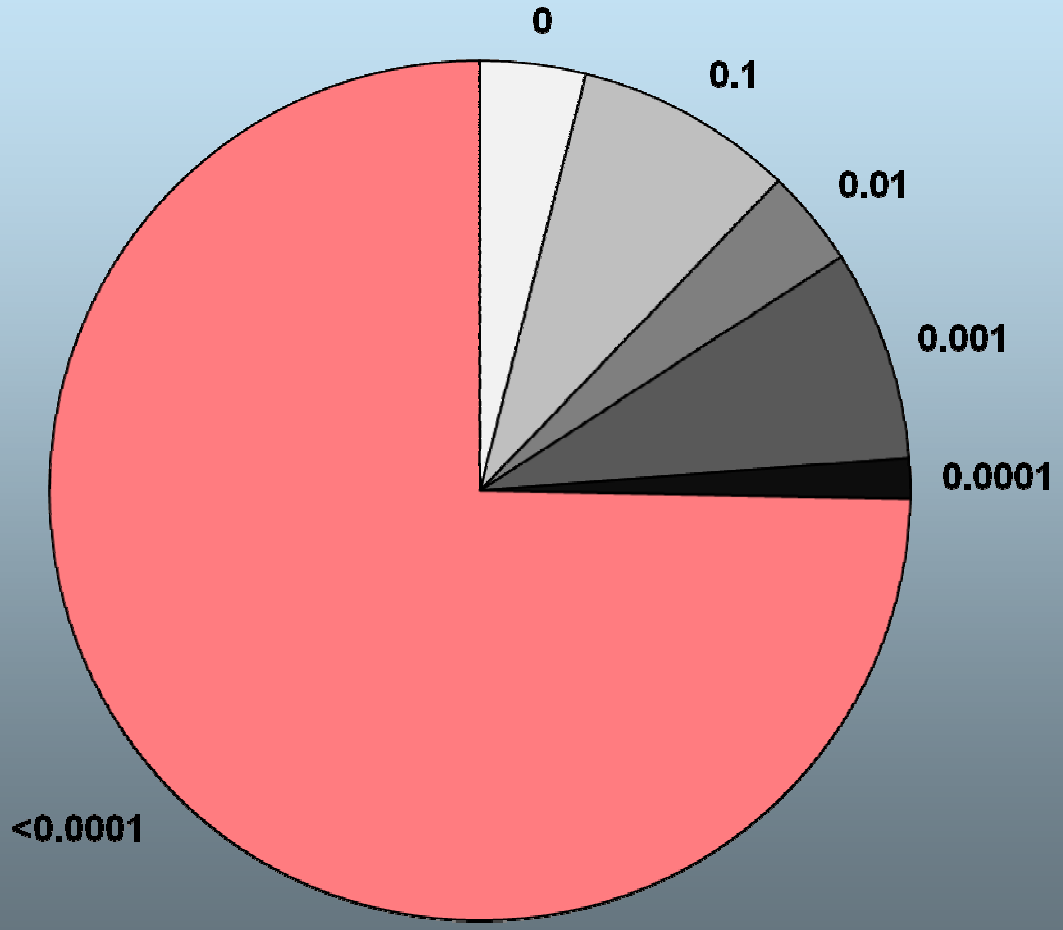
Hypothesis: Genetic tests are done at equivalent rates across Australia.

Rejected with p-value <0.0001 for 75% of tests



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The scope of the challenge

Theory

~20,000 different genes.
~100,000 different proteins or gene products.
~1,000,000 differences between any two people.
~10% of all core medical articles deal with genetics.

Practice

Hundreds of gene tests currently offered.
Assay volume *per gene* varies by factor of 10,000.
Assay volume *per lab* varies by factor of 10,000.
Marked differences in rate of testing across the nation.
Quality, training, accreditation, education, ethics, funding.

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The scope of the challenge

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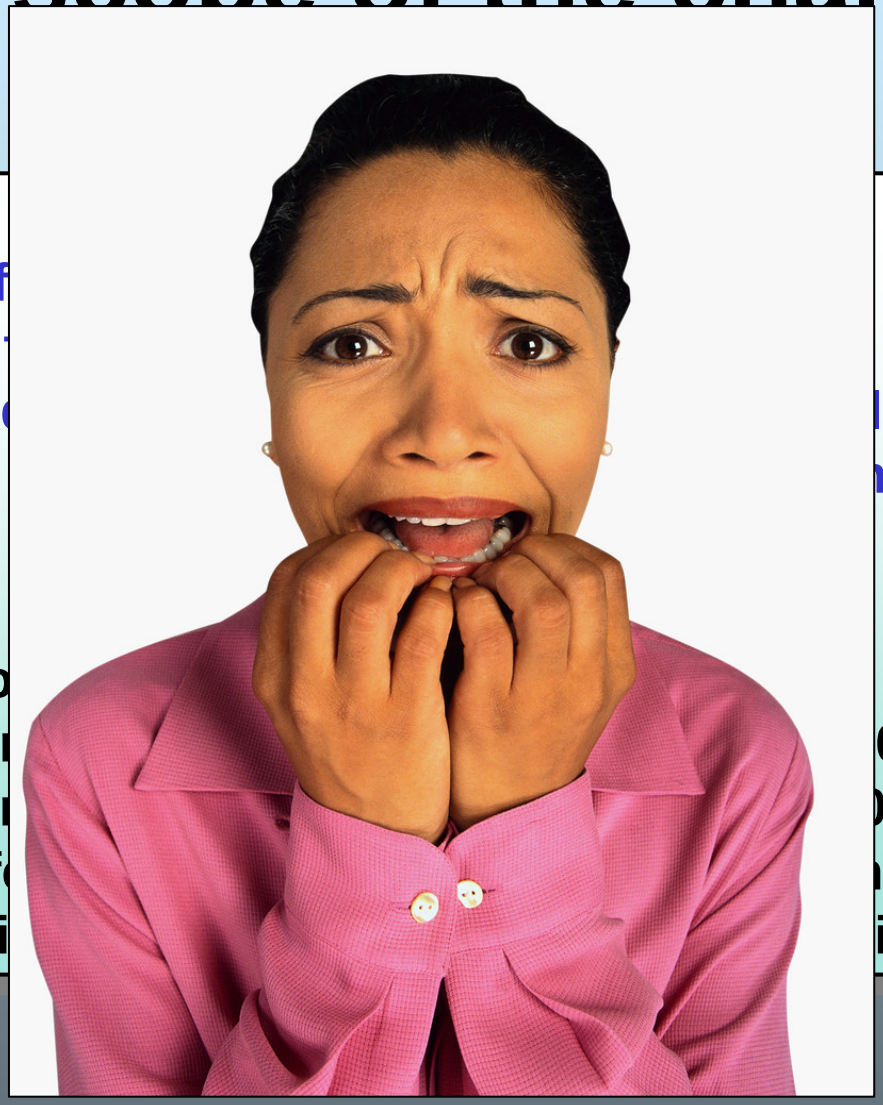

Managing the challenge

Lab numbers *i.e. training, implementation, QA etc.*

Assay vol. *i.e. edu'n ethics costs*

Assays per gene	1-2 labs	3-6 labs	7-10 labs
Low vol. <100			
Mod vol. 100-3,000			
High vol. >3,000			

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Banding by assay volume …

… makes sense re

- Genetic test diversity

Number of gene tests
by assays per gene & labs

Total Assays	Assays per gene	1-2 labs	3-6 labs	7-10 labs	Total Genes
4%	Low vol. <100	299	34	nil	76%
45%	Mod vol. 100-3,000	27	48	16	21%
51%	High vol. >3,000	7	3	1	3%

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Banding by assay volume …

… makes sense re

- Genetic test diversity
- Test complexity

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Proportion of genes sequenced
by assays per gene & labs

Assays per gene	1-2 labs	3-6 labs	7-10 labs
Low vol. <100	96%	79%	
Mod vol. 100-3,000	100%	55%	66%
High vol. >3,000	100%	75%	14%

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Banding by assay volume …

… makes sense re

- Genetic test diversity
- Test complexity
- Quality management

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Proportion of genes with ext QA
by assays per gene & labs

Assays per gene	1-2 labs	3-6 labs	7-10 labs
Low vol. <100	3%	6%	
Mod vol. 100-3,000	33%	54%	69%
High vol. >3,000	100%	100%	100%

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Banding by assay volume …

… makes sense re

- Genetic test diversity
- Test complexity
- Quality management
- Accreditation

Proportion of accredited tests
by assays per gene & labs

Assays per gene	1-2 labs	3-6 labs	7-10 labs
Low vol. <100	87%	100%	
Mod vol. 100-3,000	96%	100%	100%
High vol. >3,000	100%	100%	100%

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Banding by assay volume …

… makes sense re

- Genetic test diversity
- Test complexity
- Quality management
- Accreditation
- Assessing access & utilisation

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% of genes with equivalent testing rates across States
by assays per gene & labs

Assays per gene	1-2 labs	3-6 labs	7-10 labs
Low vol. <100	26%	32%	
Mod vol. 100-3,000	12%	17%	31%
High vol. >3,000	0%	0%	100%

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Banding by assay volume …

… makes sense re

- Genetic test diversity
- Test complexity
- Quality management
- Accreditation
- Assessing access & utilisation
- Ethical complexity

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Ethical issues
by assays per gene & labs

Assays per gene	1-2 labs	3-6 labs	7-10 labs	Genes
Low vol. <100	Rare diseases, few referrers *e.g. geneticists*			333
Mod vol. 100-3,000	Moderate demand, more referrers *e.g. specialists*			91
High vol. >3,000	High demand, common practice *e.g. GPs*			11

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Banding by assay volume …

… makes sense re

- **Genetic test diversity**
- **Test complexity**
- **Quality management**
- **Accreditation**
- **Assessing access & utilisation**
- **Ethical complexity**

…but must be based on utility and projected volumes

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Take home messages

- **Enormous diversity in labs, tests, and volume.**
- **Survey raises issues of**
  - **demand (education, access)**
  - **supply (training, funding)**
  - **implementation (managed, audited)**
  - **funding (State, Federal, patient, pharma).**
- **Stratification by "true" assay volume could assist in planning managed implementation of tests.**
- **Need a coordinated & collaborative effort by requestors, providers, and funders.**

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With thanks to

- RCPA for encouragement and support
- Quality Use of Pathology Program for funding
- Fellow pathologists and medical scientists in RCPA and HGSA for advice.
- THANK YOU to lab colleagues for the data.

Survey report now available from RCPA.

Web-list of tests and labs available at www.genetictesting.rcpa.edu.au

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