

# 20 Years of Clinical Cancer Genetic Testing in a Defined Island Population

## Bermuda: A Translational Implementation Model

### Cohort Summary (2006 – Present)

## 513 Bermudians Tested

99 dominant pathogenic variants identified across 17 cancer susceptibility genes

69% 28 BRCA1 40 BRCA2 identified variants

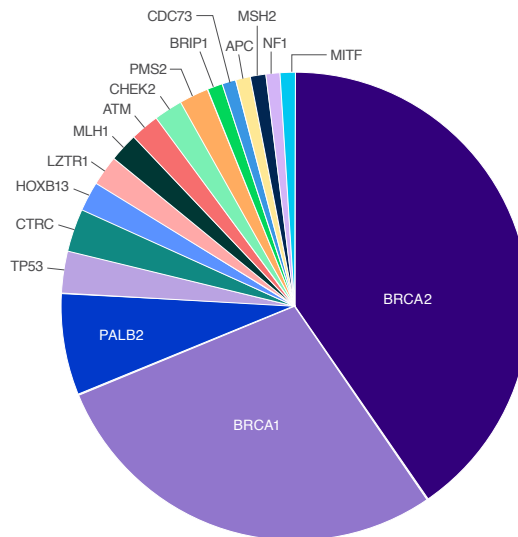
### Variant Spectrum

Pathogenic variants identified in:

BRCA1, BRCA2, PALB2, TP53, CHEK2, ATM, MLH1, MSH2, PMS2, APC, BRIP1, CDC73, HOXB13, LZTR1, NFI, MITF, CTSC

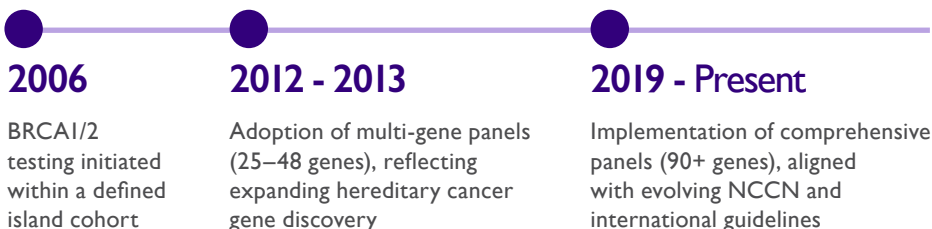
Founder mutations documented. Ancestral origins identifiable across UK, Canada, USA, Azores, and Middle East lineages.

Century-of-arrival traceable in multiple cases (1800s–2000s), reflecting island population structure and founder effects.



### Evolution of Testing & Clinical Complexity

Genetic testing in Bermuda has expanded in step with international standards and emerging gene validity evidence:



Over time, this expansion has increased variant spectrum complexity, carrier identification, and long-term clinical management requirements.

Bermuda Cancer and Health Centre  
Bermuda Cancer Genetics & Risk Assessment Clinic (Established 2006)  
Bermuda Hereditary Cancer Clinic

### Translational Infrastructure

Bermuda operates a sustained two-clinic implementation model:

#### Cancer Genetics & Risk Assessment Clinic

- Identifies high-risk individuals
- Applies guideline-based genetic testing

#### Hereditary Cancer Clinic

- Develops structured, risk-adapted management pathways
- Provides longitudinal carrier surveillance
- Facilitates cascade testing across families
- Functions as a national hereditary cancer resource
- Genetic findings initiate long-term clinical governance, not episodic testing.

### Relevance to Global Genomics

Bermuda provides:

- A defined population cohort with deep ancestral linkage
- Documented founder mutation mapping
- Longitudinal implementation over 20 years
- Integrated national-level clinical follow-up

This model demonstrates how genomic discovery can be translated into sustained, structured cancer care within a small jurisdiction.

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