PEDIGREE

Pathology, Epidemiology, DNA, Informatics and Genetics:
A Research Enabling Enterprise

What the resource is, how it is used, & opportunities for future use

Breast Cancer

Carmel Apicella
Senior Research Fellow
Centre for Epidemiology and Biostatistics
Melbourne School of Population and Global Health
The University of Melbourne

PEDIGREE

Pathology, Epidemiology, DNA, Informatics and Genetics:
A Research Enabling Enterprise

Resource ABCFR

Baseline 1992 to 2000

- 1600 women with breast cancer (VCR and NSW cancer registry)
- 1000 women without breast cancer (electoral roll)
- 9,300 adult relatives (both male and female) of these women
- 400 women of Jewish descent
- 500 individual (70 families) clinic-based
- 500 individuals (65 twin pairs) ATR
- 8000 blood samples
- ? Other biospecimens

Community Recruitment >2000

- <40 at diagnosis (VCR and Register4)
- <40 at mammogram (Register4)
- 850 new participants

PEDIGREE

Pathology, Epidemiology, DNA, Informatics and Genetics:
A Research Enabling Enterprise

Resource MCCS

Baseline 1990 to 1994 community-based (cohort)

- 41000 people
- 24000 women
- ? Blood samples
- ? Other biospecimen
- Baseline Questionnaires
- 40-69 at recruitment
- Anglo-Celtic / Greek / Italian
- Anthropometric measurements / diet

PEDIGREE

Pathology, Epidemiology, DNA, Informatics and Genetics:
A Research Enabling Enterprise

Resource Follow-up

ABCFR Follow-up

- Participant Vital Status and Cancers
- Exposures update
- New recruitment within families
- More blood samples
- Family history for other participants (no longer proband-centric)
- Expand recruitment of younger (adult) generation
- Obtain more mammograms ?

MCCS Follow-up (cohort)

- VCR cancers and death
- NDI
- Obtain mammograms for more incident cases and matched controls?

PEDIGREE

Pathology, Epidemiology, DNA, Informatics and Genetics:
A Research Enabling Enterprise

Resource Summary

- **1600 women - cohort with breast cancer at baseline**
  - ❑ **200 incident breast cancers**
- **5000 women – cohort without breast cancer at baseline**
  - ❑ **200 incident breast cancers**
- **24000 women – cohort without breast cancer at baseline**
  - ❑ **1210 incident breast cancers**

How Used

- Prospective validation of breast cancer risk prediction models
- Improve breast cancer risk prediction models by adding SNP data
- Survival of affected cohort by receptors
- Gene-environment interactions with FRP
- Contribute to consortia like BCAC (e.g. assoc snps with br ca risk)
- GWAS for br ca risk loci
- Find genes (Palb2…)

Future Use

- Risk for Relatives by adding mammographic density of cases

- Breast cancer risk prediction models including mammographic density

- Methylation

…