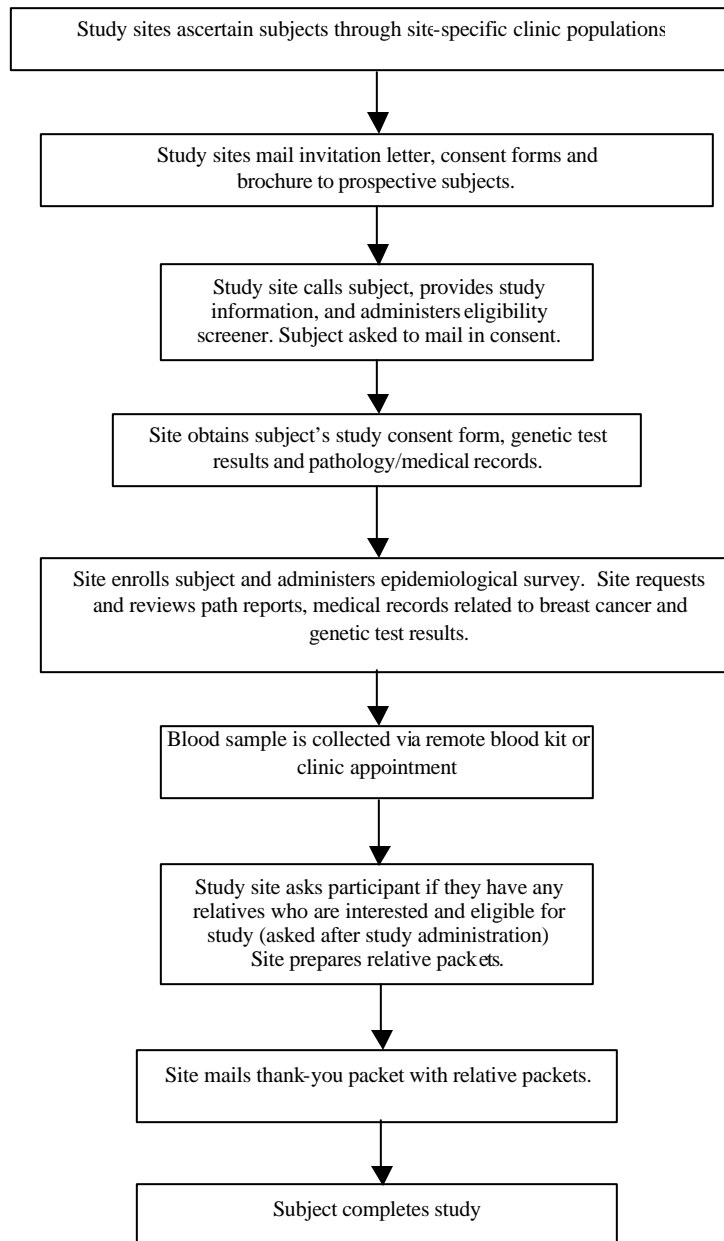


# GENETIC MODIFIERS OF BRCA1 AND BRCA2 (GEMS)

<b>Principal Investigators:</b>	Joellen Schildkraut, Ph.D.	<b>Accrual Start Date:</b>	4/2004
<b>Study Statistician:</b>	Edwin Iversen, Ph.D.	<b>Accrual End Date (Projected):</b>	11/2007
<b>Project Coordinator:</b>	Sydnee Crankshaw, MPA	<b>Study End Date (Projected):</b>	11/2008
<b>Statistical Center Coordinator:</b>	Amanda Tweed	<b>Data Cut-off Date for this Report:</b>	1/15/2005
<b>Data Management:</b>	Duke University		
<b>Statistical Support:</b>	MGH Statistical Coordinating Center		
<b>Participating Groups:</b>	CGN, SPORE		

**Goal:** A study of the genetic and environmental modifiers of breast and ovarian cancer risk associated with BRCA1 and BRCA2 mutations

## Schema for Prospective Recruitment:



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## Study Objectives

- 1) Enroll approximately 1000 female patients diagnosed with breast cancer who have tested either positive or negative for a BRCA1 and/or BRCA2 mutation in a ratio of 1 positive to 2 negatives for BRCA 1 & 2 carrier status.
- 2) Assess gene-gene interactions between BRCA1 & 2 and polymorphisms in DNA damage and repair genes.
- 3) Assess previously reported interactions between hormonally related genetic and environmental factors and BRCA1 and BRCA2 in a much larger dataset than in prior reports.
- 4) Enroll unaffected BRCA1 and BRCA2 mutation carriers to lay the foundation for future prospective studies.

## Eligibility Criteria

- Female  $\geq$  20 years old
- Type of breast cancer and confirmation of breast cancer
  - LCIS, DCIS or invasive
  - Verified with first primary pathology report(s)
- BRCA 1/2 testing (full sequencing, Jewish Panel or family mutation)
  - Been tested for BRCA1 or BRCA2 before or within 36 months (after) first primary breast cancer diagnosis.
  - Copy of the test result report for submission to Duke
- Biospecimen
  - Available to be sent to Duke (10 ml of blood or 5-20  $\mu$ g of DNA or  $3 \times 10^6$  cells)
  - Collected anytime prior to 1<sup>st</sup> primary cancer diagnosis date or within 36 months after that diagnosis
- Survey Data
  - Must have been collected within the 6 year range between 36 months prior to 36 months after the 1<sup>st</sup> primary breast cancer diagnosis
- Age matching
  - Non carriers should be matched to carriers in a 2:1 ratio based on age of diagnosis. Specifically, for each BRCA positive enrolled, 2 BRCA negatives with an age of diagnosis within  $\pm$  4 years of that positive's age of diagnosis must be enrolled.

## Participating Sites

### CGN Sites

Duke

Georgetown

Johns Hopkins

UT Southwestern

UNC-Chapel Hill

University of Pennsylvania

University of California – Irvine

### Non-CGN sites

Moffitt Cancer Institute

Vanderbilt University

Dana Farber Cancer Institute

## Accrual

As of January 15, 2005, 204 participants were enrolled.