

# Participate in a Breast Cancer Genetics Consortium



**Confluence** is an international research project—a consortium of consortia—that aims to build a large collaborative platform for **germline genetic studies of breast cancer** through the confluence of existing and new genome-wide genotyping data. It is funded by the U.S. National Cancer Institute (NCI), and coordinated by the Division of Cancer Epidemiology and Genetics (DCEG) at NCI.

## Goals

Conduct a large, multi-ancestry genome-wide association study (GWAS) to:

- Discover variants for breast cancer risk overall and by subtype
- Develop multi-ancestry polygenic risk scores for personalized risk assessment
- Discover variants for breast cancer survival, pharmacogenomics, and second cancers

## Target Size



>300,000  
Breast Cancer Cases



>300,000  
Controls

## What Studies Can Participate

Case-control, case series, or clinical trials with:

- Invasive or *in situ* breast cancer cases (female or male)
- Genome-wide genotyping data or germline DNA for genotyping
- Ethics approval and consent for germline genetic testing

## Data and Sample Request to Participate

- Germline DNA (blood or buccal): 500-1000ng by dsDNA quantification (e.g. PicoGreen)
- Genotype/sequencing data: genome-wide or mutation testing (if available)
- Phenotype data: age, pathology, treatment/toxicity, risk factors (if available)

## How to Participate

- Through collaborating Consortia (see next page) **OR**
- Collaborating directly with the NCI Coordinating Center at DCEG

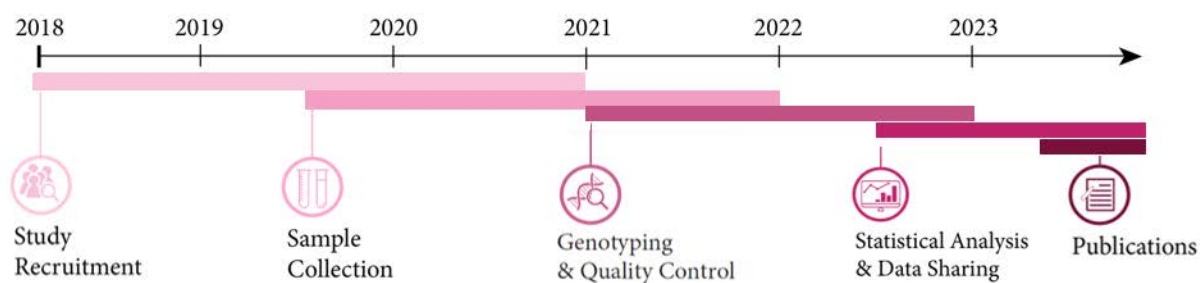
## Costs Covered by Confluence

- Data coordinating centers for study and data management
- Shipment of samples and materials (plates/tubes)
- DNA extractions at DCEG/NCI (if needed)
- Genotyping at DCEG/NCI (USA) or Cambridge University (UK) laboratories
- Return of left-over DNA (if available and requested)
- Return of genotype data to studies
- Cloud-based Collaborative Data Platform for data access and analyses

## Data Sharing Principles

- Broad data access through a secure, Cloud-based Collaborative Data Platform
- Data ownership stays with studies
- Data governed by Data Access Coordinating Committees and Data Use Agreements between data centers (NCI or Collaborating Consortia) and study institutions
- Sharing of expertise, data, and analysis code
- Co-leadership in collaborative analyses and publications
- Leverage infrastructure for additional initiatives
- Compliance with NIH Genomic Data Sharing Policy for new genotyping  
<https://osp.od.nih.gov/scientific-sharing/genomic-data-sharing/>

## Timeline



## Collaborating Breast Cancer Consortia

- Breast Cancer Association Consortium (BCAC)
- African Ancestry Breast Cancer Genetic Study (AABCGS)
- Latin America Genomics Breast Cancer Consortium (LAGENO-BC)
- Asia Breast Cancer Consortium (ABCC)
- Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA)
- Evidence-based Network for the Interpretation of Germline Mutant Alleles (ENIGMA)
- BRCA Refined Analysis of Sequence Tests: Risk of Penetrance (BRA-STRAP)
- Male Breast Cancer GWAS Consortium

# Interested in Participating?

Email: [ConfluenceProject@nih.gov](mailto:ConfluenceProject@nih.gov)

Visit: [dceg.cancer.gov/confluence](https://dceg.cancer.gov/confluence)