



**Susan G. Komen**

**Research Grants – Fiscal Year 2015**

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**Translating breast cancer genomics in the clinic**

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**Lead Organization:** University of Chicago

**Grant Mechanism:** Komen Scholars

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**Public Abstract:**

The unacceptable consequence of the marginalization of Africans and African Americans in science, reflected by their nearly complete exclusion from discovery research, has been the delayed delivery of important scientific advances - including technologies and treatments – to already vulnerable minority populations. Ironically, these are the populations most likely to benefit from these breakthroughs in science. As we know, black women die at a disproportionately higher rate from breast cancer than white women in the United States. In West Africa - the founder population of most African Americans - and for that matter in all sub-Saharan Africa, breast cancer is almost always fatal. In our study in Nigeria, we have found the majority of women presenting with a breast cancer diagnosis to be under 50 years of age and in the advanced stages of the disease. In another study from Barbados, a historically-isolated group with strong African ancestral origins, the reported incidence and mortality rates of breast cancer is the highest in the Caribbean. The significantly higher incidence of early-onset breast cancer in women of African ancestry is likely due to a correspondingly high prevalence of pertinent genetic risk factors.

To advance the field we need to further examine the specific etiology of breast cancer and its subtypes and the only way to do that is to collect more data. In our Nigerian Breast Cancer Study, we have previously recruited 1233 cases and 1101 controls in Nigeria. The University of Chicago Breast Cancer Study has recruited over 600 African American cases and 600 controls. Our goal now is to replicate our initial findings of risk factors associated with specific breast cancer molecular, or aggressive basal-like subtypes, in a larger cohort. Needed is a large-scale epidemiologic study in Ibadan, Nigeria from which we can use state-of-the-art Next Generation sequencing, genotyping, and computational biology to analyze its data. Through funding provided by the Susan G. Komen Foundation, we hope to expand our Nigerian Breast Cancer Study to collect 1500 additional cases and 1500 controls. This will help us delve further into the impact of genetic and non-



genetic factors on breast cancer risk and its molecular subtypes. To date, we have made gains in the field through developing fruitful collaborations with other investigators studying African American and African Barbadian women for comparative analyses across geographic boundaries. We have a unique opportunity to study the contribution of genetic factors to the high incidence of early onset breast cancer and overrepresentation of triple negative breast cancer in the African Diaspora. With improved access to relatively inexpensive high throughput technologies, we are now in a position to complete a comprehensive genetic analysis in a large cohort of women of African ancestry from across the globe, with well documented phenotypes. With increased numbers, we can develop a high-risk screening protocol so that individuals identified as carriers of deleterious mutations in breast cancer susceptibility genes can be enrolled in a longitudinal follow up to examine whether the outcomes can be vastly improved through family-based interventions. Funds from the Susan G. Komen Foundation will be used to enhance our recruitment and to translate findings from the genetic analysis of this cohort into meaningful interventions that hold the most promise for reducing the high mortality associated with early onset breast cancer both here and abroad.

