**A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer.**

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**Source**

Department of Preventive Medicine, Keck School of Medicine, University of Southern California/Norris Comprehensive Cancer Center, Los Angeles, USA. haiman@usc.edu

**Abstract**

Estrogen receptor (ER)-negative breast cancer shows a higher incidence in women of African ancestry compared to women of European ancestry. In search of common risk alleles for ER-negative breast cancer, we combined genome-wide association study (GWAS) data from women of African ancestry (1,004 ER-negative cases and 2,745 controls) and European ancestry (1,718 ER-negative cases and 3,670 controls), with replication testing conducted in an additional 2,292 ER-negative cases and 16,901 controls of European ancestry. We identified a common risk variant for ER-negative breast cancer at the TERT-CLPTM1L locus on chromosome 5p15 (rs10069690: per-allele odds ratio (OR) = 1.18 per allele, P = 1.0 × 10(-10)). The variant was also significantly associated with triple-negative (ER-negative, progesterone receptor (PR)-negative and human epidermal growth factor-2 (HER2)-negative) breast cancer (OR = 1.25, P = 1.1 × 10(-9)), particularly in younger women (<50 years of age) (OR = 1.48, P = 1.9 × 10(-9)). Our results identify a genetic locus associated with estrogen receptor negative breast cancer subtypes in multiple populations.