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GWAS of age at menarche and age at natural menopause

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Menarche (the first menstruation) and menopause are two significant events in a woman's life signaling the beginning and the end of reproduction. The timing of menarche and menopause varies substantially between individuals. Both age at menarche and age at natural menopause are complex traits that are influenced by genetic factors. Genome-wide association studies (GWAS) have been successful in uncovering new genetic determinants for these traits. Our initial GWAS of 17,438 women of European ancestry have identified two loci (6q16.3 and 9q31.2) for age at menarche, and four loci (20p12.3, 19q13.42, 5q35.2 and 6p24.2) for age at natural menopause. Further Meta-analysis of 32 GWAS including 87,802 women of European ancestry have identified more than 30 novel genetic loci for age at menarche and 17 for age at natural menopause. These newly identified loci implicate the biological pathways that may regulate these two traits: energy homeostasis and hormonal regulation for age at menarche, and DNA repair and immune functions for age at natural menopause. Most recently, we conducted an expanded meta-analysis of GWAS of 182,416 women of European descent that revealed 106 loci for age at menarche including multiple imprinting loci, implicating retinoic acid signaling and gama-aminobutyric acid-signalling as new mechanisms that regulate pubertal timing. These findings have expanded our knowledge of the underlying physiology and biological regulation of these traits. Insights into the genetic factors influencing the timing of menarche and natural menopause might shed light on normal reproductive function and the prevention of the diseases associated with these two traits.

Biography

Chunyan He received her ScD in Epidemiology from Harvard School of Public Health in 2009. She is an Assistant Professor in Department of Epidemiology, Indiana University Richard M. Fairbanks School of Public Health and a member of the IU Simon Cancer Center. She has published more than 25 papers in reputed journals, including three in Natural Genetics. She also serves as an editorial board member of four respected journals.

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