Researchers discover genetic links to rosacea

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Today marked the publication of the first ever genome-wide association study of rosacea, a common and incurable skin disorder. Led by Dr. Anne Lynn S. Chang of Stanford University's School of Medicine, and co-authored by 23andMe, the study is the first to identify genetic factors for this condition.

Rosacea (pronounced roh-ZAY-sha) is estimated to affect more than 16 million people in the United States alone1. Symptoms typically include redness, visible blood vessels, and pimple-like sores on the skin of the central face, and many experience stinging, burning, or increased sensitivity over the affected skin. Because rosacea affects facial appearance, it can also have a psychological impact on those who suffer from it. In surveys by the National Rosacea Society, more than 76 percent of rosacea patients said their condition had lowered their self-confidence and self-esteem.

To help better understand the genetics of rosacea, researchers at Stanford University and 23andMe studied the data of more than 46,000 23andMe customers consented for research. The study, published in the *Journal of Investigative Dermatology*, found two genetic variants strongly associated with the disease among people of European ancestry.

Further, the study uncovered that the genetic variants, or single nucleotide polymorphisms (SNPs), found to be strongly associated with rosacea are in or near the HLA-DRA and BTNL2 genes, which are associated with other diseases, including diabetes and celiac disease.

The genome-wide association study was broken into two parts: discovery and validation. First, data voluntarily submitted by 22,000 23andMe customers was examined. More than 2,600 customers reported having received a rosacea diagnosis from a physician. The remainder of the study participants did not have the condition and were treated as controls. To validate findings from this initial group, 23andMe researchers then tested the identified SNPs with a separate group of 29,000 consented 23andMe customers (3,000 rosacea patients, 26,000 controls). The researchers were able to confirm the same association with rosacea.

"This is another example of how 23andMe can help in researching common yet poorly understood diseases," said Joyce Tung, Ph.D., 23andMe's director of research and a co-author of the paper. "The study also speaks to the power of large data sets in studying and identifying genetic associations."

In addition to the genome-wide association study, the research included obtaining skin biopsies from six individuals with rosacea and showed that both HLA-DRA and BTNL2 proteins can be found in the skin of people with rosacea. This preliminary work hints toward the biological relevance of HLA-DRA and BTNL2 in rosacea.

More information: Assessment of the Genetic Basis of Rosacea by Genome-Wide Association Study, *Journal of Investigative Dermatology*, 2015.

Provided by 23andMe