MedicalXpress

23andMe identifies 5 significant genetic associations for hypothyroidism

April 9 2012

Using its unique online research platform, 23andMe, a leading personal genetics company, has found five significant genetic associations for hypothyroidism in the largest known genome-wide association study of hypothyroidism conducted to date. The details of the study are now available online in the journal *PLoS ONE*.

"With nearly 90 percent of our 125,000 customers participating in our online research, 23andMe is making crowd-sourced science a reality," stated 23andMe CEO and co-founder Anne Wojcicki. "Our online research platform continues to advance research faster and more cost effectively than traditional research models," added Wojcicki.

Of the five significant associations reported in this study, three are known to be involved in other autoimmune diseases. These include rs6679677 near PTPN22, rs3184504 in SH2B3, and rs2517532 in the HLA class I region. The other two associations reported here are rs4915077 near VAV3 and rs925489 near FOXE1, a gene involved in thyroid development. In addition, 23andMe observed associations with two loci marginally associated with hypothyroidism that have been linked to Thyroid Stimulating Hormone (TSH) levels, PDE8B and CAPZB as well as another gene previously associated with hypothyroidism and autoimmune disease (CTLA4).

"These genetic associations contribute significantly to understanding the underlying biology of hypothyroidism, which impacts five percent of the general population," remarked 23andMe Medical Director and study co-

author Dr. Uta Francke. "Identification of associations with other autoimmune disorders opens up potential pathways of treatment."

Upon identification of those novel genetic associations for hypothyroidism also associated with autoimmune diseases, the 23andMe research team looked for any additional single-nucleotide polymorphisms (SNPs) shared with other autoimmune diseases, investigating a list of 107 SNPs that were studied across seven autoimmune diseases. Among this list, only the CTLA4, PTPN22, and SH2B3 loci show significant association with hypothyroidism.

According to 23andMe Principle Scientist and Lead Author Nicholas Eriksson, Ph.D., "These findings shed new light on the biology of hypothyroidism, showing that genes involved with both thyroid function and immune response impact this disease. This demonstrates our ability to harness the power of the enormous 23andMe genetic database to further medical research.

This study investigated 3,736 individuals with hypothyroidism as well as 35,546 controls, all drawn from the more than 100,000 23andMe customers who have consented to participate in research efforts. Cases included individuals who have been diagnosed with hypothyroidism, have elevated TSH levels, or are taking thyroid hormone replacement medication.

These results continue to validate 23andMe's methodology in combining self-reported data on phenotypes gathered via web-based questionnaires and genotypic data derived from self-collected saliva samples. 23andMe had previously published its first proof-of-concept results in the journal *PLoS Genetics* in June 2010 which reported novel associations for unusual traits such as asparagus anosmia and photic sneeze reflex and replicated associations for other common genetic traits, and recently published the discovery of two novel genetic associations for Parkinson's

disease in *PLoS Genetics*, and presented a major replication study of over 180 genetic associations in the journal PLoS ONE in August 2011.

Provided by 23andMe Inc.