Researchers report new gene associated with thyroid levels

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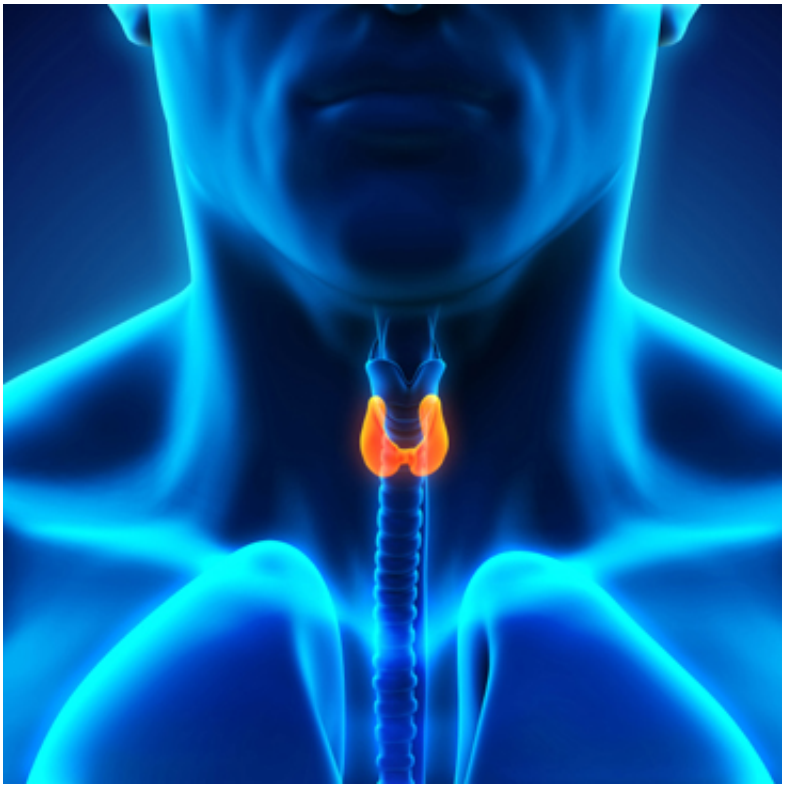

Thyroid hormones have important and diverse roles in human health and regulate metabolic rate. Thyroid disease is common (affecting 5-10 per cent of the population) and synthetic thyroid hormones are one of the commonest drug therapies prescribed worldwide. A new study, published in *Nature Communications* involving University of Bristol

academics, reports a new gene called SYN2 associated with thyroid levels.

Researchers found the SYN2 gene plays an important role in the control of thyroid stimulating hormone. The study also reports a separate, rare, genetic variant present in only four individuals per 1,000 people that can cause thyroxine levels in the blood to be elevated. Although thyroid hormones are essential for childhood development and maintaining adult health, the genetic control of these important hormones is poorly understood.

Data collected from around 4,000 people in the UK and cohorts in Europe and Australia enabled scientists to discover genes and mechanisms affecting the thyroid.

Dr Peter Taylor from the Institute of Molecular and Experimental Medicine at Cardiff University, said: "Thanks to the detailed genetic data available through a whole genome sequencing study, the UK10K project, we identified rare variants associated with thyroid hormone levels which could not have been detected in earlier studies."

The whole genomes which formed the basis of the study come from two important groups of people who have been studied for many years - the TwinsUK Adult Twin Registry and probably the most well studied adults in the world, and the University of Bristol's ALSPAC (Avon Longitudinal Study of Parents and Children) studies. The data for each group include extensive descriptions of their health and their development. Additional participants were from Busselton, Australia and Sardinia and Val Borbera, Italy.

Professor Scott Wilson, the study's lead author, said: "This whole genome sequence data has enabled us to identify that both common genetic variants with modest effects and rarer genetic variants with

larger effects determine an individual's thyroid status."

Professor Tim Spector, who leads the TwinsUK study at King's College London said, "We are fortunate to have such excellent collections of clinical research material provided by altruistic volunteers to help in medical research." More than 16,000 volunteers have contributed to this research across several countries. The base data comes from the largest genome-sequencing project so far completed. "It's humbling to see the generosity of participants in these studies. The success comes from combining the largest genome sequencing projects performed to date with the most well characterised population samples in the world."

This is one of the first in a series of UK10K studies using whole genome sequence data and clinical information to identify genetic variants which influence health right across the frequency spectrum

Dr Nicholas Timpson, one of the co-authors from the Medical Research Council (MRC) Integrative Epidemiology Unit at the University of Bristol said: "This work is another example of how extending gene studies to include whole genome sequencing can identify new clinically informative variants and enhance our understanding of key biological processes. The UK10K project has been essential to this endeavour and we are now beginning to realise its potential."

More information: "Whole-genome sequence-based analysis of thyroid function." *Nature Communications* 6, Article number: 5681 DOI: 10.1038/ncomms6681

Provided by University of Bristol