Researchers disclose genome-wide variations in secondary structure of human DNA

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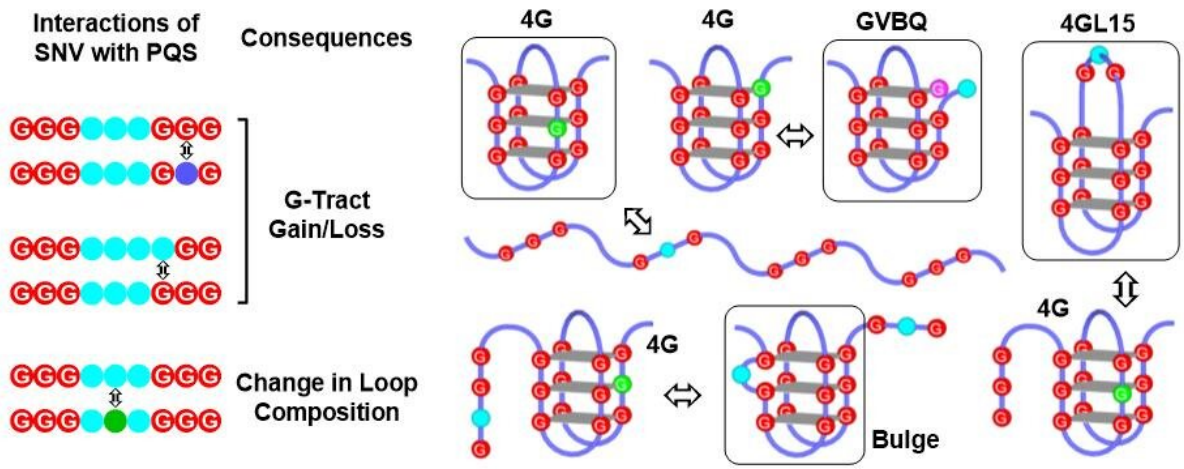

Potential G-quadruplex structure changes that can be induced by an SNV. Credit: IOZ

A recent study led by Dr. Tan Zheng from the Institute of Zoology (IOZ) of the Chinese Academy of Sciences (CAS) revealed that single nucleotide variations (SNVs) in human genomes interact with DNA motifs that can form four-stranded secondary structures called G-quadruplexes (G4s).

Such interactions result in millions of potential changes in secondary structures in the DNA of human genomes. In particular, most of these

changes occur within genes and are enriched near transcription start sites, implying they can affect gene regulation.

The study was published in *PNAS* on May 18.

The genome of each individual varies. Single nucleotide variation (SNV), a common type of variation, represents a difference in a single nucleotide in human genomes.

As the most studied genetic variations, SNVs are associated with some important traits of individuals, such as susceptibility to disease, drug response, medical outcome, and other health-related phenotypes.

Previously, research on SNV has mostly focused on the consequence of the change in an individual nucleotide in a one-dimensional context, for example, induction of change in amino acid in a protein if the SNV falls into a protein-coding region.

In this study, the researchers investigated the interaction between SNVs and G4s in a structural perspective in the whole human genome and revealed that the structural changes caused by an SNV can lead to a significant alteration in the expression of the host gene.

The new findings on the genome-wide structural variation in the human genomes provide an important addition to the mechanisms of how an SNV would influence the function of DNA.

"This will help us understand genetic variation and physiological consequences from a structural perspective. The structural variations should also represent a unique class of drug targets for individualized medicine, health assessment, and drug development," said Dr. Tan.

More information: Jia-yuan Gong et al, G-quadruplex structural

variations in human genome associated with single-nucleotide variations and their impact on gene activity, *Proceedings of the National Academy of Sciences* (2021). DOI: 10.1073/pnas.2013230118