Mediation Analyses of Ultraviolet, Air Pollution, and Structural Variations using the Taiwan Biobank

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Abstract: Structural variation is a DNA region that shows changes in copy number, sequence orientation or chromosomal location. Previous studies have suggested a link between air pollution and genetic variation in animal experiments and longitudinal studies, but the sample size is rather limited. It is imperative that a population-based study is conducted to document the potential hazard of environmental exposures such as air pollution and ultraviolet to the human genome and health. The Taiwan Biobank has been collecting biological specimens and conducts the whole-genome sequencing in order to build the reference genome of the Taiwanese population. In this study, we aim to characterize the causal relationship between ultraviolet, air pollution and structural variations. We applied a mediation model to describe the influence of ultraviolet toward structural variants through air pollution. The preliminary results showed a strong effect from ultraviolet to structural variants mediated by air pollution. Validation studies are needed to confirm this interesting finding.