TANDEM REPEATS AND PHENOTYPIC VARIATION

Microsatellites and minisatellites naturally bring to mind forensic genetics or diseases caused by their expansion. Their variability stems from their inherent propensity for changes, far more frequent than point mutations. In a review article in TIG, D.G. King (1) examines them from a different perspective, as indicated by the title of the first paragraph: “Tandem repeat polymorphisms influence phenotypic variation”. We are in the realm of evolvability. The impact of tandem repeat variations may be subtle on their own. However, when acting together, their significance can become substantial. The authors draw a comparison: "*Just as tuning knobs offer musical instruments fine, reversible adjustments, tandem repeats similarly provide a rich foundation for reversible, incremental mutation effects.*"

A similar article has been published by Lamkin and Gymrek in Nature Review Genetics (2).

These two papers highlight a crucial point regarding "missing heritability" in Genome Wide Association (GWA) studies, specifically noting the absence of tandem repeat variation. They stress that this variability must be considered, especially given its direct role in phenotypic variation.

1. https://www.cell.com/trends/genetics/abstract/S0168-9525(24)00175-6?_returnURL=https%3A%2F%2Flinkinghub.elsevier.com%2Fretrieve%2Fpii%2FS0168952524001756%3Fshowall%3Dtrue
2. https://www.nature.com/articles/s41576-024-00736-8