AN ATLAS OF VARIANT EFFECTS

Deciphering the biological implications of the very high number of human genetic variants, spanning both coding and regulatory regions, has proven to be a challenging task. Often, researchers resort to scouring previous reports for insights into variant consequences, as functional experimental data remains extremely rare. A consortium of some universities has proposed a groundbreaking initiative[1] to tackle this challenge head-on.

Their project, aptly named Multiplexed Assays of Variant Effect (MAVE), aims to comprehensively characterize the variants present in all genes and regulatory elements within the human genome. By utilizing multiplexed assays, MAVE endeavors to create a comprehensive atlas of variant effects, shedding light on their functional implications. This ambitious endeavor has the potential to revolutionize our understanding of genetics, revolutionize precision medicine, and significantly enhance the utility of genomics for diagnosing and treating disease.

Note: The recently published post titled "PRIMATE EVOLUTION AND HUMAN DISEASES" (end of July) perfectly complements the content of this post, offering valuable additional insights into the broader context of genetic variants and human health.

1- https://genomebiology.biomedcentral.com/articles/10.1186/s13059-023-02986-x