

Genetic variations in connection

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Propositions

Accompanying the thesis

Genetic variations in connections: understanding the effects of Single Nucleotide Polymorphisms in their biological context

By Elisa Cirillo

1. With the transitivity function of the BridgeDb identifier mapping framework, the user can map all the way from variant to gene to protein, enabling multi-omics analysis for various biomedical applications in different tools [chapter 3].
2. The downloadable genetic reference networks of SNPs associated with obesity, resulting from chapter 5, combine SNPs, genes, and pathways and can be used to study obesity and to develop personalized treatments.
3. *ANGPTL8* co-expression network analysis coupled with pathways analysis aided in the identification of the genes associated with the *ANGPTL8* gene regulation [chapter 6].
4. Pathway-based analysis tools for GWAS data, need to use dedicated statistical and improved visualization strategies that can combine genetic data with other omics data [chapter 2].
5. The need to develop reproducible workflows consisting of combinations of multiple connected bioinformatics tools to answer research questions is challenging even with more interoperable data coming available.
6. Epigenome-wide association studies (EWAS) are the missing link for the understanding of the regulation of complex traits.
7. Network approaches for the interpretation of complex biological data are useful when the researcher finds creative ways to highlight important aspects of the networks and consequences of the interactions.
8. Exploring and improving the ways to perform bioinformatic analyses in order to interpret results, is a key to not waste public money and to support innovation [valorisation].
9. Small changes in the genetic code can work in synergy, affecting the dance of life, via the intricate web of interacting gene products.