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CONVERGING MODELS FOR TRANSCRIPTOME STUDIES OF HUMAN DISEASES

THE CASE OF OCULOPHARYNGEAL MUSCULAR DYSTROPHY

1. The central role of transcriptional regulation, together with the comprehensive nature of current RNA expression profiling techniques, makes transcriptome data effective in modelling of biological responses. *This Thesis*
2. Phenotypic data is often treated as discretised entities whereas the most vital pieces of information are the quantitative differences and changes in phenotypes. *This Thesis*
3. Extensive phenotyping and standardization of phenotypic information aids the mechanistic understanding of rare diseases. *This Thesis*
4. In the study of a rare event such as OPMD, the identification of molecular features that are shared between varieties of biological systems uncovers insights on a broader spectrum of conditions and disorders. *This Thesis*
5. Converging loops between theoretical modelling and experimentation help to understand the dynamics of biological networks and processes. *This Thesis*
6. As non-linearity is common in biology, it is crucial to evaluate all potentially interesting relationships, independent of their form. *D. N. Reshef et al., Science 334, 1518 (2011)*
7. Integration of genomics, transcriptomics and phenomics data is a powerful approach for the characterization of correlative and causal relationships. *D. Houle et al., Nat Rev Genet. 11, 855 (2010)*
8. Consensus and precision in biological interpretation can be reached through data integration. *J. B. Tenenbaum et al., Science 331, 1279 (2011)*
9. Systematic noise due to high or low gene expression is controlled by environmental and genetic factors. *T. L. To, N. Maheshri, Science 327, 1142 (2010)*; *A. Bar-Even et al., Nat Genet. 38, 636 (2006)*; *J. R. Newman et al., Nature 441, 840 (2006)*
10. On the ordinary view of each species having been independently created, we gain no scientific explanation. *Charles Darwin*
11. Scientific knowledge is in perpetual evolution; it finds itself changed from one day to the next. *Jean Piaget*
12. You cannot depend on your eyes when your imagination is out of focus. *Mark Twain*