Cumming School of Medicine

Evolution of variable mutative organisms
Summary of Literature

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1 Project Outline

A project proposal may or may not appear here. It depends on how private the material should be.

2 References Discussion

This section is intended for me to write notes connecting the material in the references with observations and comments.

References

[1] ; on behalf of the ACMG Laboratory Quality Assurance Committee, S. Richards, N. Aziz, S. Bale, D. Bick, S. Das, J. Gastier-Foster, W. W. Grody, M. Hegde, E. Lyon, E. Spector, K. Voelkerding, and H. L. Rehm, "Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology," *Genetics in Medicine*, vol. 17, no. 5, pp. 405–423, may 2015. [Online]. Available: http://www.nature.com/articles/gim201530

[2] S. Balsamo and A. Marin, "Separable solutions for Markov processes in random environments," *European Journal of Operational Research*, vol. 229, no. 2, pp. 391–403, sep 2013. [Online]. Available: https://linkinghub.elsevier.com/retrieve/pii/S0377221713002191

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[3] E. Domingo and C. Perales, "Viral quasispecies," *PLOS Genetics*, vol. 15, no. 10, p. e1008271, oct 2019. [Online]. Available: https://dx.plos.org/10.1371/journal.pgen.1008271

A comprehensive review of support for Quasi-Species theory. This paper covers he hsitorical basis and gives a comprehensive coverage of evidence for viral QS. It also discusses the following topics, Error-prone replication and mutant spectra, phenotypic reservoir, adaptive parameters, intra-mutant spectrum quasispecies memory, sequence space, population bottle neck, biological constranits, connections with viral pathogenesis, and QS and long term evolution.

[4] J. Frazer, P. Notin, M. Dias, A. Gomez, J. K. Min, K. Brock, Y. Gal, and D. S. Marks, "Disease variant prediction with deep generative models of evolutionary data," *Nature*, vol. 599, no. 7883, pp. 91–95, nov 2021. [Online]. Available: https://www.nature.com/articles/s41586-021-04043-8

This paper covers a neural net work that uses the amino acid sequence of differnt protiens to produce a evolutionary index. The evolutionary index is then used to produce a distribution of of differnt sequences and make a prediction of B̈enign, unknowen, and Pathogenic. The distribution allows for an estimate of the confidence of the prediction. They used unsupervised learning and were able to achive an avg AUC = 0.92

[5] P. G. Harrison, "Reversed processes, product forms and a non-product form," *Linear Algebra and its Applications*, vol. 386, pp. 359–381, jul 2004. [Online]. Available: https://linkinghub.elsevier.com/retrieve/pii/S0024379504001089

NOT CLEAR YET.

[6] E. C. Holmes and A. Moya, "Is the Quasispecies Concept Relevant to RNA Viruses?" *Journal of Virology*, vol. 76, no. 1, pp. 460–462, jan 2002. [Online]. Available: https://journals.asm.org/doi/10.1128/JVI.76.1.460-462.2002

This is a letter to the editor, discussing the issues of applying quasi-species to viral RNA. It starts by critisizing the lack of formal definitions of QS. This letter ardresses, critisims (lack of def, lack of drift), and evidence (strong theoretic basis?) it makes arguments but many seem flimsy now.

[7] A. Moya, E. C. Holmes, and G.-C. Fernando, "The population genetics and evolutionary epidemiology of RNA viruses," *Nature Reviews Microbiology*, vol. 2, no. 4, pp. 279–288, apr 2004. [Online]. Available: http://www.nature.com/articles/nrmicro863

This paper reviews viral RNA mechanisms and some quantifications around the variables of mutation. Then it discusses how studies are structred for wet lab research of mutation and evolution studies. Helps to give a large overview of the topic of viral RNA without discussing Quasi-Species.

[8] on behalf of the ClinGen Sequence Variant Interpretation Working Group (ClinGen SVI), S. V. Tavtigian, M. S. Greenblatt, S. M. Harrison, R. L. Nussbaum, S. A. Prabhu, K. M. Boucher, and L. G. Biesecker, "Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework," *Genetics in Medicine*, vol. 20, no. 9, pp. 1054–1060, sep 2018. [Online]. Available: http://www.nature.com/articles/gim2017210

This paper uses naive bayes method to develop a classifier for pathogeneity of a genetic varient. This method is okay, it is not very strong, but is laying the ground work for future studies. It does a good job of relating the ACMG guidelines to a mathematical framework.

[9] C. O. Wilke, "Quasispecies theory in the context of population genetics," *BMC Evolutionary Biology*, vol. 5, no. 1, p. 44, dec 2005. [Online]. Available: https://bmcevolbiol.biomedcentral.com/articles/10.1186/1471-2148-5-44

This paper is wonderful, it describes and relates the equations from quasi-species and the conscepts from evolutionary theory. It shows how the QS equations can be derived from the principals mutation-selection equations.