Commentary Article
Volume 5:1, 2021

Journal of Genetics and Genomes

ISSN: 2684-4567 **Open Access**

Commentary on ORTHOSCOPE: A Web Tool for Genome Comparisons

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Abstract

The recent rapid growth of genomic data has made it possible to identify genes having the same inferred function, even among distantly related species, such as humans and invertebrates. Assessing evolutionary relationships provides a way to interpolate or extrapolate gene attributes among species [1]. For such evolutionary analyses, we developed a web tool called ORTHOSCOPE [2]. By uploading gene sequences of interest and by selecting species genomes from >450 bilaterians (Figure 1), users can infer their functions and copy numbers, according to results reported by ORTHOSCOPE in the form of gene trees. That is, ORTHOSCOPE can estimate the evolutionary history of a gene with one click.

Keywords: ORTHOSCOPE • Web tool • Genome • Gene tree • Species tree • Bilaterians

Case Study 1: Presence or Absence of Genes

Many studies involving genome comparisons focus only on the presence of genes belonging to selected species or lineages. In addition to the presence of genes in a lineage, ORTHOSCOPE can also evaluate their absence by selecting several representative species. For example, users can explore the evidence of horizontal gene transfer by showing the presence or absence of genes among lineages. Using Orthosocpe, we showed that the prokaryotic cellulose synthase gene (*CesA*) was horizontally transferred into the genome of a tunicate ancestor from a bacterium [3]. Tunicates are the only metazoans that can synthesize cellulose, a biological function associated with bacteria and plants, but not animals. Also, ORTHOSCOPE can be applied to such enigmas in human genomes. The possibility of horizontally acquired genes has been debated since publication of the first human genomes [4].

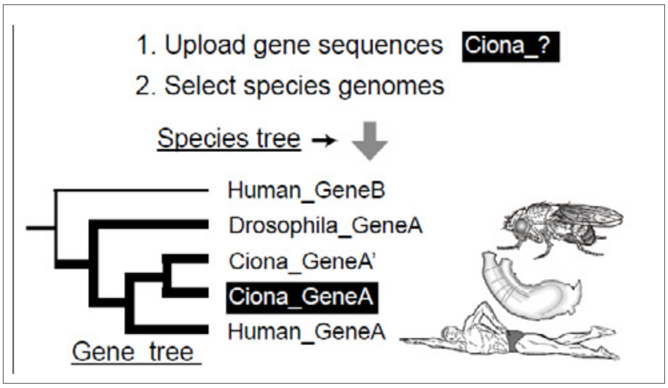

Figure 1. An overview of an ORTHOSCOPE analysis. This gene tree example indicates that the uploaded *Ciona* gene shares a function with *Drosophila* GeneA and human GeneA, and has a lineage-specific duplicate.

Case Study 2: Gene Copy Numbers

ORTHOSCOPE users can determine the number of gene copies of a species. For this purpose, ORTHOSCOPE employs a genome-scale protein-coding gene database (coding and amino acid sequence data sets) constructed for each species using only the longest sequence, when transcript variants exist for single locus. By examining 48 fish species with full genome sequences, using ORTHOSCOPE, Ishikawa et al. [5] found that multiple freshwater ray-finned fishes show a convergent increase in copies of a single gene (*Fads2*, fatty acid desaturase 2). This convergence suggests the importance of *Fads2* gene in colonization of freshwater. Also, ORTHOSCOPE can be applied to find a solution to Peto's paradox: contrary to expectation, there is little relationship between cancer incidence and the number of cells in an organism. In fact, recent studies of the African elephant genome suggest that multiple copies of tumor suppressor gene *P53* hold the answer [6].

Case Study 3: Evolutionary Relationships of Genes

ORTHOSCOPE can be used to select candidate genes for future directions in stem-cell or medical studies. The GLIS family transcription factors, GLIS1 and GLIS3, potentiate generation of induced pluripotent stem cells (iPSCs), although another GLIS family member, GLIS2, suppresses cell reprograming. Using ORTHOSCOPE, Yasuoka et al [7] showed that GLIS1 and GLIS3 originated during vertebrate whole genome duplication, whereas GLIS2 is a sister group to GLIS1/3. This study highlights the importance of carefully considering evolutionary relationships among genes without preconceptions or misconceptions stemming from classical gene names. It clearly indicates that as the first step, future reprograming studies should focus on GLIS1/3 rather than on GLIS2.

Conclusion

The web tool, ORTHOSCOPE, ensures the same environment for genome comparisons. For example, users can reproduce the same analysis used in a published paper. Thus, ORTHOSCOPE can be a standard tool for genome comparative studies.

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Received date: 24 December, 2020; **Accepted date:** 07 January, 2021; **Published date:** 14 January, 2021

References

1. Fernández R, Gabaldon T, Dessimoz C. (2020) Orthology: definitions, inference, and impact on species phylogeny inference. In Scornavacca C, Delsuc F, Galtier N, editors, Phylogenetics in the genomic era, chapter No. 2.4, pp. 2.4:1–2.4:14. No commercial publisher | Authors open access book.
2. Inoue J, Satoh N. (2019) ORTHOSCOPE: An automatic web tool for phylogenetically inferring bilaterian orthogroups with user-selected taxa. Mol Biol Evol 36: 621-631.
3. Inoue J, Nakashima K, Satoh N. (2019) ORTHOSCOPE analysis reveals the presence of the cellulose synthase gene in all tunicate genomes but not in other animal genomes. Genes (Basel) 10: 294.
4. Boto L, Pineda M, Pineda R. (2019) Potential impacts of horizontal gene transfer on human health and physiology and how anthropogenic activity can affect it. FEBS J 286: 3959-3967.
5. Ishikawa A, Kabeya N, Ikeya K, Kakioka R et al. (2019) A key metabolic gene for recurrent freshwater colonization and radiation in fishes. Science 364: 886-889.
6. Ma S, Gladyshev VN. (2017) Molecular signatures of longevity: Insights from cross-species comparative studies. Semin Cell Dev Biol 70: 190-203.
7. Yasuoka Y, Matsumoto M, Yagi K, Okazaki Y. (2020) Evolutionary history of GLIS Genes illuminates their roles in cell reprograming and ciliogenesis. Mol Biol Evol 37: 100-109.