

Commentary



Cite this article: Tong C. 2023 Convergent genomics and Arctic adaptation of ruminants. *Proc. R. Soc. B* **290**: 20232448. <https://doi.org/10.1098/rspb.2023.2448>

Received: 1 November 2023

Accepted: 4 December 2023

Subject Category:

Genetics and genomics

Subject Areas:

evolution, genomics

Author for correspondence:

Chao Tong

e-mail: tongchao1990@gmail.com

Convergent genomics and Arctic adaptation of ruminants

Chao Tong

School of Life Sciences, Arizona State University, Tempe, AZ 85287, USA

CT, 0000-0001-5202-5507

Understanding how organisms respond or adapt to extreme environments including the Arctic is a fundamental challenge in ecology and evolutionary biology. The Arctic presents an environment that is not only harsh but also extremely variable, characterized by long, cold, dark winters and short, relatively warmer summers. Such conditions require that the native fauna possess a range of physiological, morphological and behavioural adaptations for survival. Among the most fascinating examples of Arctic adaptation are the ruminants, such as muskoxen (*Ovibos moschatus*) and reindeer (*Rangifer tarandus*), that have evolved specifically to survive in the harsh Arctic environments. Recent studies have elucidated the bases of morphological, physiological or behavioural adaptation in Arctic animals, but the genetic basis of Arctic adaptation remains largely unclear. Li *et al.* [1] used a combination of genomics, molecular evolution and functional work to provide a comprehensive understanding of the mechanisms and evolutionary processes underlying convergent Arctic adaptation in ruminants.

Convergent evolution refers to the process by which unrelated or distantly related species independently evolve similar traits or adaptations as a result of having to adapt to similar environments [2]. This phenomenon of convergent phenotypic evolution serves as a powerful framework to uncover the genetic underpinnings of adaptations to specific environments, as these phenotypic convergences commonly emerge due to parallel selective pressures. With the increasing accessibility of genomic datasets of diverse species, a growing body of recent studies on convergent phenotypic evolution has unveiled remarkable genomic similarities across distinct lineages [3–9]. Convergent phenotypic evolution is hypothesized to be associated with various genetic mechanisms of genomic convergences. Many comparative genomic studies have identified convergent amino acid substitution sites in a set of candidate genes, as well as across entire genomes, revealing that distant species often exhibit parallel genetic alterations at specific loci, probably fulfilling analogous functional roles (figure 1*a*). For instance, a recent comparative genomics study between bamboo-eating giant and red pandas identified convergent amino acid substitutions in two limb development genes, *DYNC2H1* and *PCNT*. Specifically, one of the two convergent amino acid sites is R3128K in *DYNC2H1*, and the amino acid K only occurs in giant and red pandas [7]. Extensive studies aimed at identifying molecular evolutionary patterns linked to convergent phenotypic evolution have yielded different levels of genomic convergences. These include the synchronized modulation in the non-synonymous to synonymous substitution ratio (dN/dS), manifesting as either acceleration or deceleration. The shift in rate of molecular evolution involves focal foreground taxa experiencing increasing dN/dS ratios compared to background taxa, indicating acceleration. Conversely, the decrease in dN/dS ratios in foreground taxa relative to background taxa suggests deceleration (figure 1*b*) [3,10]; the observation of concerted shifts in the intensity of natural selection—either relaxation or intensification (figure 1*b*). The degree to which shifts in the distributions of dN/dS ratio across individual genes or whole genomes can be caused by relaxation (weakening of both purifying selection and positive selection, towards neutrality) versus intensification (strengthening of both purifying selection and positive selection, away from neutrality) (figure 1*b*) [3]; and the identification of common signatures of positive selection which is the dN/dS ratios

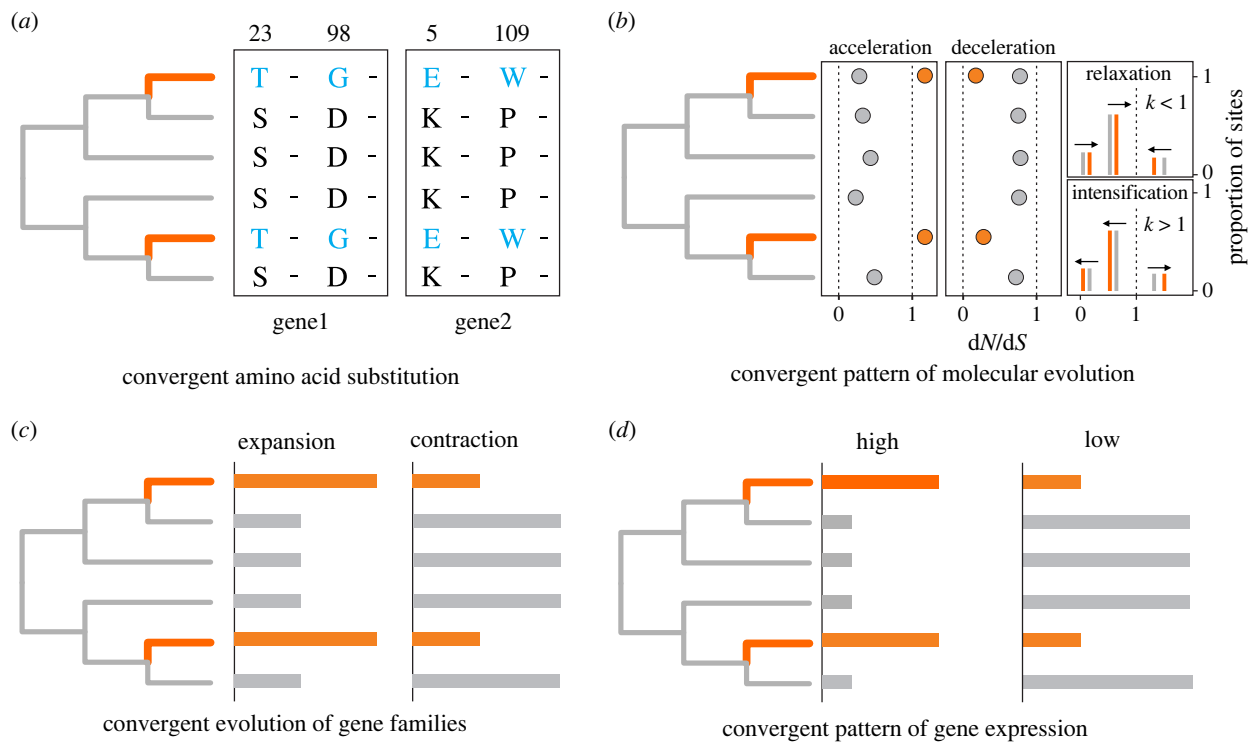


Figure 1. Types of genomic convergence. The diagram illustrates various types of genomic convergence among species sharing similar traits (orange branches, e.g. muskoxen and reindeer), contrasted with other species (grey branches) on the phylogenetic tree. Genomic convergence is categorized into four proposed types: (a) Convergent amino acid substitutions: this type involves the same amino acid substitutions occurring in both targeted species, for instance, S23T and D98G in gene1, K5E and P109W in gene2; (b) convergent pattern of molecular evolution: this encompasses parallel changes in evolutionary rates, either acceleration or deceleration, in both targeted species compared to others, and consistent patterns of relaxed or intensified selection associated with phenotypic changes; (c) convergent evolution of gene families: this type is characterized by synchronous expansion or contraction of certain gene families in the foreground species relative to background species and (d) convergent pattern of gene expression: this involves similar changes in gene expression profiles among the species with shared traits.

of specific genes or loci in focal taxa high than 1 [4,8]. Furthermore, previous studies have detected substantial convergences in gene family dynamics, notably gene loss or gain, among species exhibiting similar phenotypic traits (figure 1c) [5,9]. Complementing these findings, a growing corpus of comparative transcriptomic analyses has delineated correlations between convergent phenotypic evolution and gene expression patterns (figure 1d) [6,8,9], further substantiating the multifaceted nature of genomic convergence.

Muskoxen and reindeer represent the iconic species that have undergone convergent adaptation to Arctic extreme environments. A prior investigation into the reindeer genome had begun to reveal the genetic basis underlying Arctic adaptation [11]. However, the extent to which the Arctic milieu shapes convergent phenotypic traits, particularly within a cohort of ruminants, remains largely unknown. To address this question, the authors sequenced the muskox genome, together with the recently published reindeer genome [11] and other ruminant genomes [12]. By taking advantage of comparative genomics analysis, they identified a set of genomic convergences. These include convergent amino acid substitutions and patterns of accelerated evolutionary rates coupled with positive selection, predominantly focused on crucial genes implicated in brown adipose tissue thermogenesis and circadian rhythm regulation. In addition, they found convergent expression patterns in metabolic-related genes linked to brown adipose tissue thermogenesis, providing additional evidence of the concept of genomic convergences [1]. This paper adds to the growing list of studies that use integrative genomic approaches to elucidate

the genetic underpinnings of convergent evolution and the extent to which analogous phenotypic manifestations stem from convergent genomic alterations.

Arctic ruminants possess extraordinary adaptations for survival in extreme environments. Notably, advanced thermogenesis mechanisms enable these ruminants to efficiently conserve and use energy in Arctic environment where food can often be scarce, while unique circadian rhythms facilitated their coping with the region's extreme photoperiods [13]. In concordance with previous investigations [11,13], the authors identified distinct genomic convergences in genes involved in thermogenesis and circadian rhythm regulation. They next asked what is the functional consequences of genomic convergences during Arctic adaptation. The authors focused on one key gene, *HIF2A*, which harboured convergent amino acid substitutions in both muskoxen and reindeer, and designed functional experiments by applying mutation vector construction and co-immunoprecipitation (Co-IP). They provided putative functional evidence to support their speculation that convergence in *HIF2A* protein of both muskox and reindeer may contribute to heat transfer from brown adipose tissue to other parts of the body. This paper stood out because the specific functional assessment has often been overlooked in diverse species, despite the identification of numerous genomic convergences.

For decades, studies on genomic convergences have primarily concentrated on the evolutionary dynamics of protein-coding genes [1–4,7–9]. By contrast, recent comparative genomic studies have begun to focus on the molecular evolution of non-coding regions across the genomes of diverse species,

emphasizing the study of conserved non-coding elements (CNEs). Despite not coding for proteins, CNEs have remained remarkably unchanged across different species over millions of years of evolution [14], and are emerging as crucial components in the study of evolutionary biology [10,15]. Their primary biological functions, though not fully elucidated, are predominantly associated with the regulation of gene expression and the maintenance of genomic structural integrity. CNEs function as regulatory elements, acting as enhancers, silencers or insulators, thereby modulating gene expression spatio-temporally. Furthermore, CNEs play a critical role in developmental biology, influencing the precise expression patterns of genes during ontogeny, which is crucial for the proper morphological and functional development of organisms. The genomic datasets from Arctic ruminants, along

with those from other species exhibiting similar pronounced traits, provide the possibility for future studies to comprehensively explore patterns of convergent evolution in both protein-coding and non-coding regions within genomes, and also will elucidate in-depth genomic underpinnings of convergent adaptations.

Data accessibility. This article has no additional data.

Declaration of AI use. We have not used AI-assisted technologies in creating this article.

Authors' contributions. C.T.: writing—original draft, writing—review and editing.

Conflict of interest declaration. I declare I have no competing interests.

Funding. I received no funding for this commentary.

References

- Li M *et al.* 2023 Convergent molecular evolution of thermogenesis and circadian rhythm in Arctic ruminants. *Proc. Biol. Sci.* **290**, 20230538.
- Storz JF. 2016 Causes of molecular convergence and parallelism in protein evolution. *Nat. Rev. Genet.* **17**, 239–250. (doi:10.1038/nrg.2016.11)
- Tong C, Avilés L, Rayor LS, Mikheyev AS, Linksvayer TA. 2022 Genomic signatures of recent convergent transitions to social life in spiders. *Nat. Commun.* **13**, 6967. (doi:10.1038/s41467-022-34446-8)
- Sun Y-B, Fu T-T, Jin J-Q, Murphy RW, Hillis DM, Zhang Y-P, Che J. 2018 Species groups distributed across elevational gradients reveal convergent and continuous genetic adaptation to high elevations. *Proc. Natl Acad. Sci. USA* **115**, E10634–E10641.
- De Smet R, Adams KL, Vandepoele K, Van Montagu MCE, Maere S, Van de Peer Y. 2013 Convergent gene loss following gene and genome duplications creates single-copy families in flowering plants. *Proc. Natl Acad. Sci. USA* **110**, 2898–2903. (doi:10.1073/pnas.1300127110)
- Hart JC, Ellis NA, Eisen MB, Miller CT. 2018 Convergent evolution of gene expression in two high-toothed stickleback populations. *PLoS Genet.* **14**, e1007443. (doi:10.1371/journal.pgen.1007443)
- Hu Y *et al.* 2017 Comparative genomics reveals convergent evolution between the bamboo-eating giant and red pandas. *Proc. Natl Acad. Sci. USA* **114**, 1081–1086. (doi:10.1073/pnas.1613870114)
- Hao Y, Xiong Y, Cheng Y, Song G, Jia C, Qu Y, Lei F. 2019 Comparative transcriptomics of 3 high-altitude passerine birds and their low-altitude relatives. *Proc. Natl Acad. Sci. USA* **116**, 11 851–11 856. (doi:10.1073/pnas.1819657116)
- Xu S, Wang J, Guo Z, He Z, Shi S. 2020 Genomic convergence in the adaptation to extreme environments. *Plant Commun.* **1**, 100117. (doi:10.1016/j.xplc.2020.100117)
- Kowalczyk A, Partha R, Clark NL, Chikina M. 2020 Pan-mammalian analysis of molecular constraints underlying extended lifespan. *Elife* **9**, e51089. (doi:10.7554/eLife.51089)
- Lin Z *et al.* 2019 Biological adaptations in the Arctic cervid, the reindeer (*Rangifer tarandus*). *Science* **364**, eaav6312. (doi:10.1126/science.aav6312)
- Chen L *et al.* 2019 Large-scale ruminant genome sequencing provides insights into their evolution and distinct traits. *Science* **364**, eaav6202. (doi:10.1126/science.aav6202)
- Blix AS. 2016 Adaptations to polar life in mammals and birds. *J. Exp. Biol.* **219**, 1093–1105. (doi:10.1242/jeb.120477)
- Leybold NA, Speicher MR. 2021 Evolutionary conservation in noncoding genomic regions. *Trends Genet.* **37**, 903–918. (doi:10.1016/j.tig.2021.06.007)
- Treaster S, Deelen J, Daane JM, Murabito J, Karasik D, Harris MP. 2023 Convergent genomics of longevity in rockfishes highlights the genetics of human life span variation. *Sci. Adv.* **9**, eadd2743. (doi:10.1126/sciadv.add2743)