

# The remarkable tuatara finds its place

Rebecca N. Johnson

The genome sequence of an unusual reptile called the tuatara sheds light on the species' evolution and on conservation strategies. The work is a model of current best practice for collaborating with Indigenous communities. **See p.403**

A once-species-rich order of reptiles called the Rhynchocephalia lived across the globe during the time of the dinosaurs<sup>1,2</sup>. Just one of these species survives today: the tuatara (Fig. 1). Found only in New Zealand, tuatara are a *taonga* ('special treasure') for Maori people. The reptiles have a set of intriguing traits – including longevity and an unusual combination of bird- and reptile-like morphological features<sup>3</sup> – that have led to uncertainty over their place in the evolutionary tree. On page 403, Gemmell *et al.*<sup>4</sup> report the first whole-genome sequence for the tuatara

(*Sphenodon punctatus*). The researchers' study provides insights into the biology and evolution of this extraordinary animal.

The work is a collaboration between genomicists and Ngātiwai, the Maori *iwi* (people) who have guardianship over the tuatara populations used in this study. Even with the advances in genome-sequencing technology over the past several years, it is not possible to produce a high-quality genome sequence without access to good genetic material. The researchers obtained this only through collaboration. Ngātiwai were involved in all decision-making

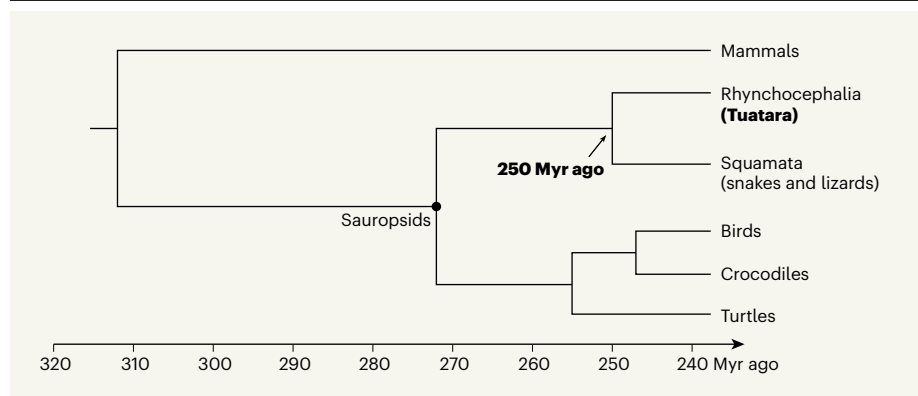
processes for this study, and are commendably listed as the paper's last authors. Gemmell *et al.* also provide a template agreement that other researchers can follow should they wish to consult with traditional guardians of other organisms. As such, the study sets a new standard for collaboration with Indigenous guardians on genomics and other scientific endeavours.

The genome produced by Gemmell and co-workers is one of the largest vertebrate genomes published so far. At more than 5 gigabases, it is about 50% larger than the human genome. To complement the genome, the authors generated gene-expression profiles for tuatara blood and embryos. They also performed a preliminary analysis of active and inactive sections of the genome, and an in-depth analysis of repeated regions. The genome represents a valuable resource for future research into a variety of topics – from the evolution of egg laying to why the once-species-rich Rhynchocephalia has only a single survivor.

One reason for sequencing genomes is to reconstruct the evolutionary tree of life; this allows a deeper understanding of how life evolved, and this knowledge can be used to tackle challenges such as biodiversity loss and climate change. Gemmell *et al.* used comparative-genomics methods to do just that.



**Figure 1** | A tuatara in New Zealand. Gemmell *et al.*<sup>4</sup> have generated a high-quality genome sequence for the tuatara (*Sphenodon punctatus*).



**Figure 2 | Refining the evolutionary tree for reptiles, birds and mammals.** This phylogenetic tree includes six branches: mammals and five branches within a clade called sauropsids, which comprises reptiles and birds. One of these, the Rhynchocephalia, has only one living member, the tuatara. Gemmell and colleagues date the divergence of the Rhynchocephalia from the Squamata to about 250 million years (Myr) ago.

They generated a phylogenetic tree for the Sauropsida (a clade that includes all modern reptiles, along with birds) by comparing the genome sequences of 27 vertebrates, including the tuatara (Fig. 2). The researchers' tree confirms a previous suggestion<sup>5</sup> that the Rhynchocephalia diverged from their closest relatives<sup>5,6</sup>, the Squamata (lizards and snakes), about 250 million years ago, during the Early Triassic period. Confirmation of such an early divergence is important for understanding the origin and evolution of the Lepidosauria, which comprises both the Rhynchocephalia and the Squamata.

Could the tuatara be a living fossil? The term, which refers to a species that has evolved extremely slowly and still retains the features of its ancient ancestors, has fallen out of favour with palaeontologists and evolutionary biologists. This is due, in part, to misuse of the term, which can arise when fossil evidence that would have reflected physical changes in a species is missing, or when researchers mistakenly assume that a lone survivor of a given lineage must have remained unchanged over evolutionary time. Tuatara have a close resemblance to their forebears from the early Mesozoic era<sup>7</sup>, between 240 million and 230 million years ago. However, there is no continuous fossil record<sup>6</sup>, making it difficult to define which traits the tuatara might share with its now-extinct ancestors.

Gemmell and colleagues' phylogenetic reconstruction indicates that the tuatara has the lowest rate of evolution of any lepidosaur described so far. These data could suggest that the tuatara is indeed a living fossil. In addition to its long generation time and low body temperature, the tuatara's slow evolution could make it particularly vulnerable to a warming climate.

The authors then analysed the tuatara's genome in more detail. On average, more than 50% of a vertebrate's genome is comprised of repetitive DNA sequences (repeat elements)<sup>8,9</sup>. In line with this figure, 64% of the

tuatara genome is repeat elements. However, the types of repeat element were a combination of mammal-like and reptile-like. This is a key finding, because the most-recent common ancestor of sauropsids was imputed to be reptile-like on the basis of genomic features found in birds and lizards, some of which have very well-characterized genomes<sup>10</sup>. By revealing unexpected, mammal-like features, the tuatara genome provides new evolutionary insights.

The researchers also found that the tuatara genome has a broader range of DNA sequences called transposons (sequences that can move from one genomic location to another) than has any other reptile, bird or mammal sequenced so far. Many of these seem to have been active recently (probably in the past

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few million years), suggesting that they still have or have recently had a role in shaping the genome. The authors suggest that the tuatara's large genome might be explained by the fact that almost one-third of it consists of duplications of DNA sequences between 1 and 400 kilobases long.

Gemmell *et al.* then compared tuatara genes associated with eyesight, smell, immunity, thermoregulation and longevity with the equivalent genes in other species. Despite being nocturnal, the tuatara is a highly visual predator, and the authors found evidence that it has retained vision-associated genes reminiscent of an ancestor that was active during the day. The species seems to have retained robust colour vision, even at low light levels – suggesting that there could be an adaptive benefit to having this trait.

In addition, tuatara seem to have a repertoire of several hundred odour receptors – similar to the number in birds, but lower than that in crocodiles or turtles. Further research is required to investigate the function of these receptors and to determine the implications of this reduced receptor repertoire for tuatara feeding and hunting. For instance, perhaps tuatara rely on their vision for hunting (like birds), rather than depending on odours and other senses (as do snakes).

Finally, there is an ongoing debate about whether there are actually two subspecies of tuatara – crucial information for conservation strategies. Because the animals are protected, the authors assessed genetic diversity among the population using samples collected over many decades. This analysis confirms that there is only one species of tuatara, despite one population (on North Brother Island in the Cook Strait) being genetically distinct from the others. The lack of current samples is not desirable for designing genetics-based conservation approaches, but, given the tuatara's longevity, any recommendations arising from the study are still likely to be valid.

Much as whole-genome sequencing has benefited human health and improved our understanding of human evolution, the sequencing of genomes of other organisms can have many benefits – especially for those organisms facing biodiversity loss caused by humans. However, for many such species, samples are not readily available. Gemmell and colleagues' work reminds us that sample collection and consultation with Indigenous people can go hand in hand to improve outcomes for both biological and cultural conservation.

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1. Evans, S. E. *Biol. Rev. Camb. Phil. Soc.* **78**, 513–551 (2003).
2. Hsiou, A. S. *et al. Sci. Rep.* **9**, 11821 (2019).
3. Gunther, A. *Phil. Trans. R. Soc. Lond.* **157**, 595–629 (1867).
4. Gemmell, N. J. *et al. Nature* **584**, 403–409 (2020).
5. Jones, M. E. H. *et al. BMC Evol. Biol.* **13**, 208 (2013).
6. Jones, M. E. H. & Cree, A. *Curr. Biol.* **22**, R986–R987 (2012).
7. Herrera-Flores, J. A., Stubbs, T. L. & Benton, M. J. *Palaeontology* **60**, 319–328 (2017).
8. Pasquesi, G. I. M. *et al. Nature Commun.* **9**, 2774 (2018).
9. Sotero-Caio, C. G., Platt, R. N. II, Suh, A. & Ray, D. A. *Genome Biol. Evol.* **9**, 161–177 (2017).
10. Zhang, G. *et al. Science* **346**, 1311–1320 (2014).

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