

## From the archive

Musings about correct scientific spelling, and an ancient Egyptian weighing balance.

### 100 years ago

May I follow Prof. Grenville Cole ... in supporting Sir Clifford Allbutt? The prefix “dino-,” as thus spelled, is ambiguous. We who know that “dinosaur” means “terrible lizard” may smile at the undergraduate and his “dinnosaur.” But how would you pronounce “Dinocystis”? Wrongly, no doubt, as I did myself until I learned that the first begetter of the name derived it from δῖνεiv, to swirl, because the rays are spirally coiled. The same for Dinocharis and Dinophysa ... [W]hat about the giant corkscrew shell from the Hastings Sand — the Dinocochlea of B. B. Woodward? That perhaps means “spiral coil”; or does it mean “monster coil”? Should it, in short, be Deinocochlea or Dinocochlea?

From *Nature* 22 December 1923

### 150 years ago

I have to thank Mr. Rodwell for calling my attention, in *NATURE* ... to the curious representation of an equal-armed Egyptian balance in a papyrus, now in the British Museum. This papyrus, which is perhaps the most beautiful in the whole collection, all the colours and lines being as bright and distinct as when originally painted, has been shown to me by Dr. Birch ... The heart of the deceased is being weighed in an equal-armed balance, and found lighter than a feather. In the papyrus, the weighing is being made in the Hall of perfect Justice, in presence of Osiris ... [W]hat Mr. Rodwell mentions as a sliding weight on one side of the beam, appears rather to be a loop or ribbon for limiting the oscillation of the beam. In the original papyrus the middle and both ends of the beam, as well as the lower part of the column, are coloured to represent polished brass, whilst the other parts of the balance are dark, as if of bronze. It should be observed that the balance beam has boxends for suspending the pans. Judging from the height of the human figures, the length of the balance beam represented is about six feet, and the height of the column ... is nearly the same.

From *Nature* 18 December 1873



For example, the authors’ ingenious dual-gate control could be used to realize synaptic plasticity in the vestibulo-ocular reflex, the mechanism that stabilizes images on the retina as the head moves<sup>8</sup>. It will be interesting to see what other models of plasticity can be expressed, such as spike-timing dependent plasticity, in which the strengthening of a synapse is dependent on the timing of stimulation<sup>9</sup>.

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## Genetics

# Indigenous diversity in Australia’s DNA tapestry

**Katrina G. Claw & Amber Nashoba**

Diverse genomic sequences might boost equity in areas such as health care. Genomic data from Indigenous Australians, shared through a community-consultation framework, aids efforts to boost genetic representation. **See p.593**

Languages and genes can tell us stories of the past. The genetic differences between Indigenous Australian communities are not well understood but are hypothesized to reflect their linguistic diversity. On page 593, Silcocks *et al.*<sup>1</sup> present findings supporting this hypothesis, showing exceptionally high genetic variation and strong population structure (patterns of genetic variation resulting from past non-mixing) in genomic comparisons of linguistically distinct communities. The authors also highlight the importance of respecting cultural perspectives and engaging Indigenous Australian peoples in genomic research and its applications.

Australia, covering 7.6 million square kilometres, is geographically larger than the European Union (4.1 million square kilometres) and smaller than the United States (9.8 million square kilometres). More than 250 Indigenous languages are spoken in Australia, and these are an essential part of Indigenous medicine, identity and ancestral connections. Genetic analysis is another way to establish connections, because geographical patterns of genetic variation correlate with linguistic variation.

Worldwide, efforts to actively involve Indigenous ancestors (and their descendent communities) and peoples in genomic research are infrequent, with some research groups being more successful than others. In the past decade there have been several frameworks and guidelines published by

Indigenous communities and stakeholders, describing current approaches and recommended improvements<sup>2–4</sup>.

Silcocks *et al.* analysed data from the largest group (cohort) of Indigenous peoples to have had its genomes sequenced and published so far, with community consultation and input. The authors investigated how communities are related, working with Indigenous Australians to analyse full genomes of previously unanalysed speakers of certain Indigenous language groups.

Indigenous Australian peoples have lived for time immemorial on remote tropical islands, on northern coasts rich in biodiversity and beauty, and on lands nestled among the red sand dunes of the Simpson Desert in the centre of the country. Silcocks and colleagues present work sequencing 159 individual genomes from Indigenous Australian communities at four such sites (Fig. 1), and compare these data with some previously sequenced<sup>5,6</sup> genomes of 60 individuals from Papua New Guinea and its associated islands. The four sites were the Tiwi islands, Galiwin’ku and Yarrabah, all in the north of the country; and Titjikala in central Australia. Writing in *Nature*, Reis *et al.*<sup>7</sup> analysed DNA from the same cohort studied by Silcocks and colleagues to look at structural variation, examining the linear order of nucleotides in a genome and alterations to this order such as inversions, duplications and deletions.

Silcocks *et al.* report some of the highest levels of previously uncharacterized genetic variation seen outside Africa, where populations have the greatest amount of genetic diversity in the world. Interestingly, the authors' findings support a genetic separation of Indigenous Australian peoples from the Indigenous inhabitants of Papua New Guinea about 47,000 years (1,636 generations) ago, with the Australian peoples of the Tiwi islands separating from other Indigenous Australians about 35,000 years (1,006 generations) ago.

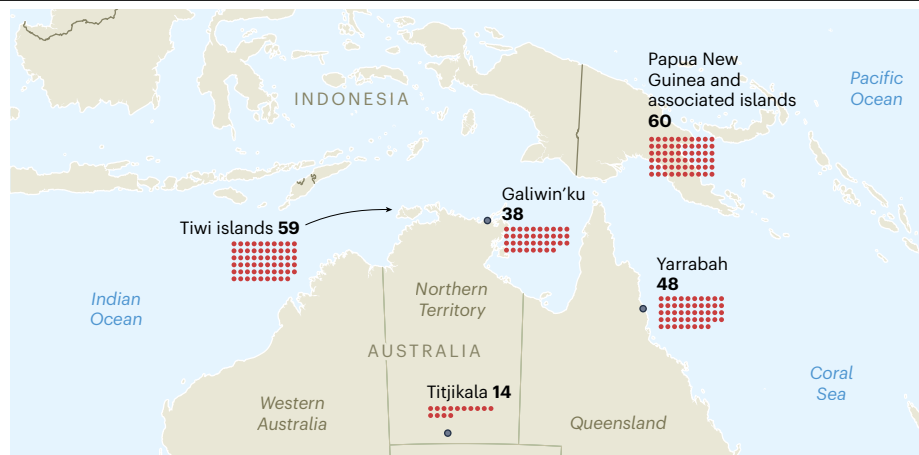
Indigenous Australians comprise extremely diverse groups with complex histories influenced by traditional territories, cultural affiliations, languages and geographical barriers. They should therefore not be treated as a single group in future studies. This diversity underscores the need to include more Indigenous Australians in genetic-variant databases and genome assemblies.

The data give support to the early inhabitation of Australia by Indigenous peoples who share a complex relationship with Papuan peoples. It is striking that genetic variation (measured by what is termed the fixation index, or  $F_{ST}$ ) between communities at Galiwin'ku and Titjikala, separated by approximately 1,400 kilometres, resembles the degree of genetic variation between peoples from Cambodia and Oroqen (in northeast China), who are 4,500 km apart.

Previous Indigenous Australian genomic studies<sup>5,8</sup> demonstrate the long history these peoples have with the land and environment, often taking us into uncharted ethical territory related to the stewardship of ancestors in museums or other historical collections. Silcocks and colleagues' work represents a major milestone and attests to the continued improvements and possibilities of community-engaged and community-based participatory research approaches in genomics.

Nearly five decades after the first sample collection and after more than a decade of consultation with Indigenous leaders, the establishment of the National Centre for Indigenous Genomics (NCIG) in Canberra in 2013 has placed a large collection of biosamples from 69 Indigenous Australian communities under Indigenous governance and custodianship, and built trust and engaged in dialogue with communities and participants. As of 2016, these ancestors have been under Indigenous-majority custodianship at the NCIG. The centre's research-ethics protocol (see [go.nature.com/3rcqf7w](https://go.nature.com/3rcqf7w)) focuses on respect, participant control of samples and data, and projects that matter to Indigenous Australian communities.

Silcocks *et al.* demonstrate that ethical and scientific gains are possible when informed consent of research participants is built into the design of a study. Community members' willingness to contribute samples for this



**Figure 1 | Locations of Indigenous populations providing samples for genomic analysis.** Silcocks *et al.*<sup>1</sup> analysed genomic data for individuals (number indicates genomes per site) from linguistically distinct Indigenous communities at four sites in Australia, and compared these with previously sequenced<sup>5,6</sup> genomes from Indigenous communities in Papua New Guinea and its associated islands. These data shed light on migrations and on the timing of when these populations separated from communities in Papua New Guinea. This genomic information might also prove a useful resource to enable more equitable health care.

research reflects the crucial collaboration fostered through the NCIG. Continuing to build trust and develop these relationships might expand community participation and enable future collaborative research. The lack of representation of Indigenous peoples in genomic data sets is not an insurmountable obstacle, and can be improved.

The authors demonstrate that inclusion improves analytical robustness and inferential accuracy, and is a crucial component of their direct findings and methodology. They report that one-quarter of the genetic variants they identified are previously unknown, overturning the idea that genomic databases contain comprehensive records of human variants. These findings have the potential to contribute to clinical diagnostics and point the way to future work to improve “genomic medicine and equitable healthcare”, as mentioned by Silcocks and colleagues.

We hope that in future work with Indigenous Australians, communities are represented in the study design in a manner that reflects the linguistic and cultural variation that is known to correspond to their genetic diversity. This emphasizes the need to include Indigenous Australians in otherwise broadly sampled variant databases and genome assemblies.

In addition, further progress can be made when all researchers attend more thoughtfully to how historical shifts in scientific terminology can powerfully address what are flagged as “objectifying and disrespectful” conventions<sup>3</sup>. Increasing awareness of how population descriptors reflect community self-determination and adherence to ethical standards should be a priority among researchers<sup>9</sup>. Terms such as consanguinity and recent inbreeding should be used with caution because of potential misunderstandings or stigma to communities, and alternative and

specific terms such as identity by descent or haplotype sharing used instead.

Although Indigenous peoples do not need scientific evidence to validate their knowledge, it is reassuring to see that Western science does sometimes catch up with and complement Indigenous knowledge systems and histories. The collective knowledge and historical memory of Indigenous communities exists in Indigenous peoples and the past is inextricably linked to the present and the future. This comprehensive study shows that community-engaged genomic research is achievable and something all researchers in this field should strive for. Community engagement, supportive and inclusive leadership and the acknowledgement of the self-determination of Indigenous people will enable genomic data collection to increase inclusion and equity for all peoples.

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