Ancient DNA as Historical Evidence

Ancient DNA and History: Origins and Development

The application of ancient DNA to the study of history has its conceptual roots in the thought of Luca Cavalli-Sforza, an Italian geneticist convinced that the analysis of the constitutive biology of modern individuals could facilitate insight into historical 'big questions' such as prospective migrations by their ancestors. In the 1980s, Cavalli-Sforza and his colleagues themselves migrated into the realm of DNA analysis, having exhausted the utility of blood types and other data sources: through studying the genetic makeup of their contemporaries, they sought to make inferences about the historical activities of their forebears.[1] For Reich, Cavalli-Sforza was a visionary but faintly tragic figure who provided the rationale behind ancient genetic research but worked in 'the dark ages of DNA', when technological limitations meant that the prospect of analysing substantial amounts of ancient DNA itself, much less sequencing the entire genome of specific ancient individuals, was thought improbable.[2]

The first attempts at studying DNA from ancient remains came in the 1980s, when material from Egyptian mummies was examined.[3] This yielded tiny amounts of genetic information about the target individual in the form of short, low-quality DNA strands, alongside microbial and fungal matter which the post-mortem breakdown of the body had introduced.[4] Each

[1] D. Reich, *Who we are and how we got here: ancient DNA and the new science of the human past* (Oxford, 2018), pp. xii-xv.
[2] *Ibid.,* p. xviii.
[3] E. Willerslev and A. Cooper, 'Ancient DNA', *Proceedings: Biological Sciences,* 272: 1558 (January 7, 2005), p. 3.
[4] M. Parks et al., 'Ancient population genomics and the study of evolution', *Philosophical Transactions: Biological Sciences,* 370: 1660 (19 January, 2015), p. 2.

strand of what little endogenous DNA was recovered then required molecular cloning before study: this was an inefficient, time-intensive and expensive process which involved researchers manually growing thousands of bacterial colonies to learn about highly degraded material.[5]

In 1985, Polymerase Chain Reaction (PCR) method enabled DNA, both ancient and modern, to be multiplied in a test-tube. PCR uses DNA 'primers' to target desired sequences of information and induce their duplication, enabling researchers to amplify specific fragments for further examination. This technique, it transpired, had its own practical problems: its sensitivity made it susceptible to contamination by modern DNA from excavators, archaeologists and laboratory workers.[6] This brought an attendant increase in experimental load by necessitating that different samples from the same individual be sent to distinct sites for the replication of results. In turn, the destructiveness of aDNA research increased, which, it has been recognised, is not without ethical implications.[7]

The 2010s saw a 'technological paradigm shift' which drastically improved the usefulness of aDNA research to historians.[8] To reduce PCR's cost to laboratories in both time and money, shotgun sequencing was increasingly used: this mobilises computing power to read bulk genetic data *en masse*, categorising it on behalf of scholars and in doing so, eliminating the need for arduous and pricey manual processing.[9] The Reich laboratory at Harvard pioneered

---

[5] S. Pääbo, 'Ancient DNA', *Scientific American*, 269: 5 (November 1993), pp. 86-88.
B. Shapiro and M. Hofreiter, 'A paleogenomic perspective on evolution and gene function: new insights from ancient DNA', *Science*, 343: 6169 (24 January, 2014), p. 1.
[6] Pääbo, 'DNA', pp. 89-90.
[7] C.D. Sarkissian et al., 'Ancient genomics', *Philosophical Transactions: Biological Sciences*, 370: 1660 (19 January, 2015), p. 1.
L. Orlando et al., 'Ancient DNA analysis', *Nature Reviews Methods Primers*, 1: 14 (2021), p. 2.
[8] E. Hagelberg, M. Hofreiter and C. Keyster, 'Introduction: Ancient DNA: the first three decades', *Philosophical Transactions: Biological Sciences*, 370: 1660 (19 January, 2015), p. 2.
[9] Reich, 'Who we are', pp. xix-xx.
J. U. Adams, 'Complex genomes: shotgun sequencing', *Nature Education*, 1 (1):186 (2008), p. 186.

the streamlining of the entire extraction and analysis process, employing robots, specialist technicians and computer scientists to serve up data for interpretation. In this way, it became possible for Reich and his team to extract whole-genome data for most remains examined: this collapsed the cost of aDNA analysis but was chiefly impactful for historians because of its implications for chronology.[10] Human DNA contains about three billion nucleic acids, three million of which, in any given individual, deviate from the species-type: because these mutations arise consistently over time, it has been noted that the number of differences between two individuals can reveal much about historical concerns such as ancient population mixtures and migrations.[11] Slatkin and Racimo draw a parallel with the cartographic feats of early-modern Europeans in terms of the extent of the expansion of our knowledge of the past which this enables; whether or not this is justifiable, it is evident that the relevance of aDNA research to historians has increased markedly in recent years - a weighing of its advantages and disadvantages is in order.[12]

Evaluating the aDNA revolution and its impact on History:

As we have seen, the general trend in the development of aDNA studies has seen this research become more precise, less time-consuming, and less financially draining with time, especially since in the 2010s. Generic inaccuracy, delay and expense, though, were not the only problems with pre-2010s aDNA research: contamination and ethics must also be considered.

---

[10] Reich, 'Who we are', p. xx.
P. Nair, 'Sequencing Ancient DNA', *Proceedings of the National Academy of Sciences*, 111: 7 (February 18, 2014), p. 2401.
[11] Reich, 'Who we are', pp. 3-4.
[12] M. Slatkin and F. Racimo, 'Ancient DNA and human history', *Proceedings of the National Academy of Sciences*, 113: 23 (7 June, 2016), pp. 6385-6386.

The problem of contamination:

The aDNA revolution has been massively impactful in its relative liberation of the field from this issue. In archaeological settings, contamination is a concern because modern DNA readily mixes with ancient DNA – not least when excavators and archaeologists themselves share similar genetic signatures with the deceased – and can corrupt datasets in consequence. The PCR method itself risked contamination in laboratory settings: when test-tubes in which multiplication had occurred were opened, imperceptible air movements effectively blew ancient DNA material onto surfaces in a way which risked the integrity of subsequent findings from that location. Authentication through the replication of results minimised this but doubled both the workload of the discipline and its (literally crushing) impact on ancient remains.[13] The use of whole-genome data has ameliorated the consequences of contamination by making it far easier to spot where modern DNA has produced aberrations in the data and to correct for this in results.[14] Though the need to maintain strict anti-contamination regimes and measures such as routine cleaning, the wearing of PPE and the maintenance of access regulation remains paramount, the reliability borne of decreased contamination risk is clearly greatly beneficial to historians.[15]

Destruction, ethical hazard and the problem of preservation:

Though worthwhile efforts at minimising the destructiveness of aDNA extraction through the identification of minimally invasive skeletal locations have been made, the fact remains that

[13] Willerslev and Cooper, 'DNA', pp. 6-7.
[14] Reich, 'Who we are', p. xx.
[15] Orlando et al., 'DNA analysis', p. 4.

it remains damaging to ancient remains. Researchers sandblast the skulls of their subject to recover the petrous pyramid in the inner ear, the most DNA-rich part of the body, or failing that, teeth or thoracic bones.[16] When biological studies use data derived from living people, their informed consent is obtained or the work is regarded as unethical and often deemed unlawful.[17] In the case of ancient individuals, this is an inherent impossibility, so an ethical 'weighing-up' of relevant considerations has been pursued. On one hand, scholars have recognised that the interests of scientific and historical understanding – including amongst general publics descended from the relevant ancient individual – emphatically side with extraction, whilst on the other, it is hard to dispute that having one's head sawn open post-mortem represents an invasive violation of bodily autonomy and the peace of the dead.[18] To resolve this question, scholars have wondered whether consultation with descendants might offer a way to provide a voice for the deceased in deliberations, but in most cases, the idea that there can be a genuine affinity in outlook between ancient and modern individuals of the same genetic composition is probably spurious.[19] This represents an unsatisfying impasse which the aDNA is yet to fully address: given the importance of ethical concerns to responsible historians, not least given the propensity for genetics to be instrumentalised to license modern barbarism against the marginalised, this is deeply problematic.[20]

The quandary above speaks to a broader risk intrinsic to historicising genetic evidence in the modern world: the threat that findings will be mobilised in favour of deterministic arguments about the propensities and capabilities of modern groups. This is particularly worrisome in

[16] C. Parker, 'A systematic investigation of human DNA preservation in medieval skeletons', *Scientific Reports*, 10 (2020), pp. 1-10.
[17] K.A. Horsburgh and F. Kaestle, 'Ancient DNA in Anthropology: methods, applications, and ethics', *American Journal of Physical Anthropology*, 119: S35 (13 December, 2002), p. 106.
[18] I. M. Kaufmann and F. J. Rühli, 'Without 'informed consent'? Ethics and ancient mummy research', *Journal of Medical Ethics*, 36: 10 (October 2010), pp. 608-610.
[19] S. Holm, 'The Privacy of Tutankhamen – Utilising the Genetic Information in Stored Tissue Samples', *Theroretical Medicine and Bioethics*, 22 (2001), pp. 440-443.
[20] Horsburgh and Kaestle, 'Ancient DNA in anthropology', p. 109.

the case of aDNA because of a preservation bias which favours the survival of material embedded in cold, DNA-preserving conditions – this has meant that the amount of aDNA evidence available tends to increase with distance from the equator. Though the analytical strides of recent years have made it easier to examine deteriorated remains from humid and tropical locales, it is easy to imagine how this could have negative implications for modern peoples if historians are not careful.[21] For instance, it is extremely likely that evidence for the northern hemisphere will reveal highly developed ancient trade routes and networks.[22] Historians must communicate to the public that this is a function of an environmentally imposed bias in the availability of evidence which does not give grounds for arguments from silence about 'isolated' or 'inactive' Global Southerners.

aDNA and History in practice:

The practicalities of aDNA research entail opportunities and problems for historians. That aDNA is a heterogenous field with considerable input from the sciences represents an opportunity for engagement across conventional disciplinary lines. However, this has led to the drowning-out of historical knowledge when, for example, aDNA articles are placed in scientific journals with restrictive word counts. Similarly, government and third-party funding for aDNA scholarship, which admittedly skews experiment-heavy, is disproportionately allocated to scientific institutions.[23] For historians themselves, acquiring the specialist scientific knowledge involved in aDNA research is a tall order given other workload demands, including teaching, writing and language acquisition. At an institutional and individual level, then, the aDNA revolution is mixed in its implications for historians.

[21] Slatkin and Racimo, 'DNA and human history', p. 6386.
[22] Orlando et al., pp. 19-20.
[23] *Ibid.*, pp. 6, 20.

Conclusion:

The potential for aDNA to inform the study of history is overwhelmingly strong. Over time, aDNA research has become more accurate, less laborious, and cheaper. The 2010s were transformative for those reasons, but equally importantly, saw technological innovation render contamination, once imagined an inescapable scourge, manageable. The ethical implications of aDNA research mean that historians should engage in the field in a morally serious and informed way. The practical facts make it necessary for historians to engage in cooperation with scholars from other disciplines, and to act as advocates for the historical method and the fruits it has already borne when they do so.

aDNA introductory reading list

General introduction and historical applications:

Reich, D., *Who we are and how we got here: ancient DNA and the new science of the human past* (Oxford, 2018), introduction and chapter 1.

Slatkin, M. and Racimo, F., 'Ancient DNA and human history', *Proceedings of the National Academy of Sciences*, 113: 23 (7 June, 2016), pp. 6380-6387.

The science, before and after technological takeoff:

Cooper, A. and Willerslev, E., 'Ancient DNA', *Proceedings: Biological Sciences*, 272: 1558 (January 7, 2005), pp. 3-16.

Orlando, L. et al., 'Ancient DNA analysis', *Nature Reviews Methods Primers*, 1: 14 (2021), pp. 1-26.

Ethics:

Kaufmann, I.M.and Rühli, F.J., 'Without 'informed consent'? Ethics and ancient mummy research', *Journal of Medical Ethics*, 36: 10 (October 2010), pp. 608-613.

Holm, S., 'The Privacy of Tutankhamen – Utilising the Genetic Information in Stored Tissue Samples', *Theroretical Medicine and Bioethics*, 22 (2001), pp. 437-449.

Bibliography

Adams, J.U., 'Complex genomes: shotgun sequencing', *Nature Education*, 1 (1):186 (2008).

Cooper, A. and Willerslev, E., 'Ancient DNA', *Proceedings: Biological Sciences*, 272: 1558 (January 7, 2005), pp. 3-16.

Hagelberg, E., Hofreiter, M. and Keyster, C., 'Introduction: Ancient DNA: the first three decades', *Philosophical Transactions: Biological Sciences*, 370: 1660 (19 January, 2015), pp. 1-6.

Holm, S., 'The Privacy of Tutankhamen – Utilising the Genetic Information in Stored Tissue Samples', *Theroretical Medicine and Bioethics*, 22 (2001), pp. 437-449.

Horsburgh, K. A. and Kaestle, F., 'Ancient DNA in anthropology: methods, applications, and ethics', *American Journal of Physical Anthropology*, 119: S35 (13 December, 2002), pp. 92-130.

Kaufmann, I.M.and Rühli, F.J., 'Without 'informed consent'? Ethics and ancient mummy research', *Journal of Medical Ethics*, 36: 10 (October 2010), pp. 608-613.

Nair, P., 'Sequencing Ancient DNA', *Proceedings of the National Academy of Sciences*, 111: 7 (February 18, 2014), p. 2401.

Orlando, L. et al., 'Ancient DNA analysis', *Nature Reviews Methods Primers*, 1: 14 (2021), pp. 1-26.

Pääbo, S., 'Ancient DNA', *Scientific American*, 269: 5 (November 1993), pp. 86-92.

Parker, C., 'A systematic investigation of human DNA preservation in medieval skeletons', *Scientific Reports*, 10 (2020), pp. 1-16.

Parks, M. et al., 'Ancient population genomics and the study of evolution', *Philosophical Transactions: Biological Sciences*, 370: 1660 (19 January, 2015), pp. 1-10.

Reich, D., *Who we are and how we got here: ancient DNA and the new science of the human past* (Oxford, 2018).

Sarkissian, C.D. et al., 'Ancient genomics', *Philosophical Transactions: Biological Sciences*, 370: 1660 (19 January, 2015), pp. 1-12.

Shapiro, B. and Hofreiter, M., 'A paleogenomic perspective on evolution and gene function: new insights from ancient DNA', *Science*, 343: 6169 (24 January, 2014), pp. 1-7.

Slatkin, M. and Racimo, F., 'Ancient DNA and human history', *Proceedings of the National Academy of Sciences*, 113: 23 (7 June, 2016), pp. 6380-6387.