**Institution: The University of Huddersfield**

**Unit of Assessment: 5 Biological Sciences**

**Title of case study: Using archaeogenetics to understand human history and ancestry**

## 1. Summary of the impact

Work by the University of Huddersfield's Archaeogenetics Research Group has been at the forefront of developing mitochondrial DNA as a tool for reconstructing the dispersal history of mankind, from a new model of the expansion of modern humans out of Africa to re-evaluations of the settlement history of Europe, Asia and the Pacific. Pivotal in the emergence of commercial genetic ancestry testing, this work generates immense public interest and creates many opportunities for broad engagement. It has provided an expert basis for TV and radio programmes, featured widely in the mainstream press and helped the Human Genetics Commission formulate guidelines for the genetic ancestry testing industry.

## 2. Underpinning research

Archaeogenetics uses genetic evidence to address questions about evolution, archaeology, history and other elements of the human past. The University of Huddersfield's Archaeogenetics Research Group, based within the Department of Biological Sciences, has played a leading role in using the non-recombining genetic marker systems – especially mitochondrial DNA (mtDNA), but also the Y chromosome – to trace the female and male lines of descent and make inferences from the distributions and diversity of genealogical lineages about settlement patterns and population dispersals. This work, which concerns humans in the main but also domesticates and commensals, is a subject of major public interest.

The Head of the Archaeogenetics Research Group, Professor Martin B. Richards (Senior Lecturer, University of Huddersfield, 2000–2004; Senior Lecturer and Professor of Archaeogenetics, University of Leeds, 2004–2011; Professor of Archaeogenetics, University of Huddersfield, 2012–), and his colleague, Dr Dougie Clarke (Senior Lecturer, University of Huddersfield, 1998–2006; Head of Biological Sciences, University of Huddersfield, 2006–), have been at the forefront of the ongoing process of establishing and developing archaeogenetic methodologies. These have been applied to many of the "big questions" about human history, such as the settlement of the world, the spread of farming and the impact of climate change. In each area Huddersfield has significantly reshaped the debate and thereby informed public understanding.

The novel phylogenetic methodologies that have emerged from this work have allowed researchers in the Archaeogenetics Research Group and their colleagues to elucidate in detail the mtDNA tree and the definition of its lineages or haplogroups. These form the basis for all subsequent discussion, including the commercial tests provided for the public by the genetic ancestry industry – particularly since the research initially focused on short control-region sequences, which were also targeted by the testing companies. In addition, new phylogeographic methodologies have made possible the interpretation of the data and allowed the research to contribute to – and in several cases revolutionise – debates concerning some of the major topics surrounding human evolution.

For example, by showing that European genetic patterns were shaped mainly by climate change at the end of the last Ice Age [2,3], these studies challenged the established view that European ancestry traces primarily to the Neolithic. They have also shown that the consensus "out of Taiwan" model for the origins of island Southeast Asians and Pacific Islanders is incorrect, with major dispersals again in the wake of the Ice Age, reaching New Guinea by about 6,000 years ago [4]. Furthermore, the research has laid the ground for the study of African mtDNA variation, identifying markers tracing the Bantu expansion into southern Africa and again identifying earlier (especially postglacial) processes [5].

Perhaps most significantly, this work has proposed a new model for the dispersal of modern humans out of Africa. It suggests there was a single expansion along the southern coast of Asia around 60,000 years ago, so refuting both a single route through northeast Africa and the Levant and the possibility of multiple dispersals [1,6]. This model has become widely recognised as the consensus since its publication in 2005.

## 3. References to the research

**Publications** (Huddersfield academics in bold):


1. Macaulay, V, **Hill, C,** Achilli, A, Rengo, C, **Clarke, D, Meehan, W, Blackburn, J,** Semino, O, Cruciani, F, Scozzari, R, Taha A, Shaari, NK, Raja, JM, Ismail, P, Zainuddin, Z, Goodwin, W, Bulbeck, D, Bandelt, H–J, Oppenheimer, S, Torroni, A, **Richards, M** (2005): Single, Rapid Coastal Settlement of Asia Revealed by Analysis of Complete Mitochondrial Genomes, *Science*, 1034-1036. doi: 10.1126/science.1109792 (Scopus 25/11/13 = 310) [work carried out as Hill's studentship project during Richards' time at Huddersfield]
2. **Richards, M,** Macaulay, V, Torroni, A, Bandelt, H–J (2002): In Search of Geographical Patterns in European mtDNA, *American Journal of Human Genetics*, 71, 1168-1174. doi:10.1086/342930 (Scopus 25/11/13 = 83)
3. Torroni, A, Bandelt, H–J, Macaulay, V, **Richards, M,** et al. (2001): A Signal, from Human mtDNA, of Postglacial Re-Colonization in Europe, *American Journal of Human Genetics*, 844-852. doi:10.1086/323485 (Scopus 25/11/13 = 161)
4. **Hill, C,** Soares, P, Mormina, M, Macaulay, V, **Clarke, D,** Blumbach, PB, Vizuete-Forster, M, Forster, P, Bulbeck, D, Oppenheimer, S, **Richards, M** (2007): A Mitochondrial Stratigraphy for Island Southeast Asia, *American Journal of Human Genetics*, 80, 29-43. doi:10.1086/510412 (Scopus 25/11/13 = 94) [work carried out as Hill's studentship project during Richards' time at Huddersfield]
5. Salas, A, **Richards, M,** De la Fe, T, Lareu, MV, Sobrino, B, Sanchez-Diz, P, Macaulay, V, Carracedo, A (2002): The Making of the African mtDNA Landscape, *American Journal of Human Genetics*, 71, 1082-1111. doi:10.1086/344348 (Scopus 25/11/13 = 276) [work mainly carried out during visit of Salas to Huddersfield in 2001]
6. Fernandes, V, Alshamali, F, Alves, M, Costa MD, Pereira JB, Cherni, L, Harich, N, Cerny, V, Soares, P, **Richards, MB,** Pereira L (2012): The Arabian Cradle: Mitochondrial Relicts of the First Steps Along the Southern Route out of Africa, *American Journal of Human Genetics*, 90, 347-355. doi:10.1016/j.ajhg.2011.12.010 (Scopus 25/11/13 = 17)


**Some research grants** [* = awarded to/funds held at Leeds, but remaining active and carried over to Richards' time at Huddersfield]:

- *Radiocarbon dating and the peopling of late glacial Europe and the Near East*. The Leverhulme Trust Research Project Grant to MBR with Dr Paul Pettitt (University of Bristol) (as Co-Investigators) and Professor Clive Gamble (University of Southampton, Principal Applicant). May 2001–April 2004. £84,185
- *Which way out of Africa?* Royal Society Research Grant to MBR. August 2003–July 2004. £10,000
- *The modern human settlement of south-east Asia.* British Academy Project Grant. Award reference: LRG–35407, to MBR. April 2003–March 2005. £20,000
- *The first stop of the modern human migration out of Africa (75,000-60,000 years ago): high-resolution characterization of informative mitochondrial DNA, Y-chromosome and autosomal markers in the Arabian Peninsula and Near East.* FCT studentship (to MBR, with Dr Luisa Pereira, IPATIMUP), December 2009–November 2013. 50,000 Euros*
- *Characterising modern human dispersals in the Greater Mediterranean by combining genetic and archaeological data.* FCT studentship (to MBR, with Dr Luisa Pereira, IPATIMUP), February 2009–January 2013. 50,000 Euros*
- *Genetic characterisation of the European settlement by modern humans in the Early Upper Palaeolithic.* FCT studentship (to MBR, with Dr Luisa Pereira, IPATIMUP), February 2009–January 2013. 50,000 Euros*
- *Complete mitochondrial DNA sequence variation in the Malay Peninsula*. Universiti Sains Malaysia funded studentship (to MBR, with Dr Stephen Chia, USM and Dr Stephen Oppenheimer, University of Oxford), February 2010–January 2014. £31,575 consumables*
- *Anchoring New Guinea in Southeast Asia and Melanesia: multidisciplinary investigations of long-term social interaction and identity formation.* Australian Research Council Project Grant, Partner Investigator MBR, with Chief Investigators Professor Mark Donohue

(Australian National University) and Dr Tim Denham (Monash University), January 2010–December 2012, £234,000 (402,000AuD)

- *Complete mtDNA variation and the modern human settlement of Southwest Asia.* Leverhulme Trust Research Project Grant (to MBR; Co-I's Dr Vincent Macaulay (University of Glasgow), Professor Antonio Torroni (University of Pavia), and Douglas Baird (University of Liverpool), September 2010-August 2013. £147,965*
- *Demography of modern humans in Southeast Asia and the Portuguese Overseas Empire Influence: a genetic approach.* FCT studentship (to MBR, with Dr Luisa Pereira & Dr Pedro Soares, IPATIMUP), January 2012–December 2015. [50,000 Euros consumable funding withdrawn due to Portuguese government cutbacks]
- *Immortalisation of ancient DNA extracts as libraries and targeted DNA-capture of whole mitochondrial genome sequences from ancient humans/hominids*. Royal Society Project Grant, to MBR and Dr Paul Brotherton, April 2012–March 2013, £15,000

## 4. Details of the impact

The work of the Archaeogenetics Research Group has significantly contributed to furthering the understanding of human history and ancestry, benefiting not just immediate peers and experts in related fields but also a range of non-specialist audiences.

Having originally provided one of its main pillars, the ARG's research continues to underpin the growing genetic ancestry testing industry. The companies operating within this sphere draw on both their published mtDNA sequences and the system of haplogroup nomenclature and details of the distributions of lineages of mtDNA, data analyses and interpretations developed by the ARG and their colleagues. This influence reaches across firms in the UK and the US, as well as the large-scale National Geographic/IBM-funded Genographic Project, and has stimulated numerous amateur genealogists to set up websites dedicated to their own mtDNA lineage or providing information for fellow enthusiasts. Dr Jim Wilson [a], who has founded several successful ancestry testing companies, has remarked: "Richards' research underpins significant parts of the mtDNA products available from all genetic ancestry testing companies, a global market with over half a million customers. From his earliest studies through to his latest work on West Asia, Africa, Europe and Far East, his discoveries of the structure of the mtDNA tree, the phylogeography of the branches and advances in dating combine to reveal the ancestral female line history of modern humans."

Conscious that the impact of such companies might not always be positive, Richards has also played a key role in providing guidance for the industry. In March 2010, having specifically requested his input, the Human Genetics Commission, which until May 2012 served as the UK government's advisory body on new developments in the field, published revised advice for the general public, warning of genetic ancestry tests' inherent limitations. In August 2010 this led to HGC publishing *A Common Framework of Principles for Direct-to-Consumer Genetic Testing Services* [b], which included guidance on genetic ancestry tests.

The ARG's expertise has impacted on society and culture through extensive media involvement, particularly in the form of advice for TV and radio programmes. The ARG has also sought to analyse and evidence the benefits of this input by surveying the new audiences it has been able to reach.

In 2009, Richards was consultant for the BBC Two series *The Incredible Human Journey* [c], which was screened in May and June that year and attracted an average of 2m viewers per episode. The programme drew especially on work in references 1–3 for its narrative. 26 out of 120 respondents in a survey carried out in October 2012/June 2013 (see below) remembered seeing the show, and more than three-quarters of these said it changed their view of human evolution. Series producer Paul Bradshaw has remarked: *"It has been clear from the public response that [the programme] stirred up wide interest in this fundamental area and helped put this deep ancestral story into the consciousness of the nation... Martin's input had a steadying and confidence-building effect on our narrative and, in turn, helped inform and enlighten our viewers."*

Richards was also consultant for the first episode of BBC One's *Andrew Marr's History of the World* [d], screened in September 2012 to an audience of 3.85m. Research on the out-of-Africa dispersal (references 1, 6) was dramatised over the first fifteen minutes of the programme, for

which Richards also checked and amended the voice-over script. Marr wrote in the *Radio Times*: "When I discovered... that every person now alive who is not a sub-Saharan African shares ancestry from a single tribe that left Africa some 70,000 years ago… well, that felt like a wonderful place to begin." 19 out of 120 survey respondents saw the programme, and almost two-thirds of these said it changed their view of human evolution. The series is now used in schools as a teaching aid. Producer/director Guy Smith has credited Richards' *"authoritative contribution"* with *"making a difficult subject accessible to a broad television audience".*

Richards was also interviewed on film and consulted for *Norwegian Roots* [e], a three-part documentary series by the Norwegian Broadcasting Company (NRK), which was shown first on NRK1 in March 2012 to a 36% audience share (one of the most viewed programmes in Norway that week) and drew on work in references 2 and 3. This led to the ARG's involvement in a production for *Newton*, NBK's children's science series, telling the story of human migrations through mtDNA. Richards was also an adviser for BBC One's *Meet the Izzards* [f], screened in March 2013 and attracting an audience of 2.6m, in which comedian Eddie Izzard used DNA to trace his ancestry out of Africa and into Europe, and for a 10-minute feature on the modern human dispersal out of Africa for an April 2011 episode of BBC One's *Bang Goes the Theory*, which typically attracts more than 3m viewers. *Meet the Izzards* assistant producer Alex Gliddon has acknowledged Richards' role as of *"crucial importance to relaying complex subjects to the wider public".* Richards has argued the case for phylogeographic analysis in a May 2013 *Guardian* Notes and Theories blog [g], correcting some common misconceptions about how the results are achieved, and has also advised on the archaeogenetic aspects of works of popular history, such as Higham and Ryan's *The Anglo-Saxon World* (YUP, June 2013).

The ARG's efforts to communicate its work to a broader audience have been further supported by a number of talks to the general public. These have included lectures and discussions at the University of Leicester (February 2008), Nottingham Science Café (March 2008), the University of Oxford (April 2011) and Otley Science Café (July 2011), with an invitation to speak at a day school entitled "Palaeolithic Archaeology: Current Scientific Investigations" at the Department for Continuing Education, University of Oxford, in October 2014. In July 2012, and again in July 2013, members of the ARG also gave a series of talks to A-level students from Greenhead College, Huddersfield, leading to enthusiastic feedback on Facebook.

In October 2012 Richards delivered another public lecture, *The Genetic Time Machine: Archaeogenetics and Human Ancestry*, to an audience of around 250 at the University of Huddersfield (delivered again in June 2013 at the Barnsley Campus). Afterwards, 41 out of 120 survey respondents "completely agreed" with the statement "The lecture told me things I didn't know about human evolution and dispersals that changed my views of this area of science"; 49 "agreed". Some 73 per cent of respondents said they were stimulated to find out more about human origins, and almost half of those who responded said the lecture changed their view on whether to take a genetic ancestry test [h].

**5. Sources to corroborate the impact**

a. Dr Jim Wilson, founder, Ethnoancestry, ScotlandsDNA, BritainsDNA, IrelandsDNA

b. Bionews report on publication of *A Common Framework of Principles for Direct-to-Consumer Genetic Testing Services*, August 9 2010 http://www.bionews.org.uk/page_68221.asp

c. Paul Bradshaw, producer, *The Incredible Human Journey*

d. Guy Smith, producer/director, *Andrew Marr's History of the World*

e. Svein Haaland, head of documentary, NRK (*Norwegian Roots)*

f. Alex Gliddon, assistant producer, *Meet the Izzards*

g. Martin Richards and Vincent Macaulay, *The Guardian* Notes and Theories, 8/4/13: http://www.theguardian.com/science/blog/2013/apr/08/unfair-genetic-ancestry-testing-astrology

h. Martin Richards, Public Lecture survey results (Excel spreadsheet, obtainable on request)