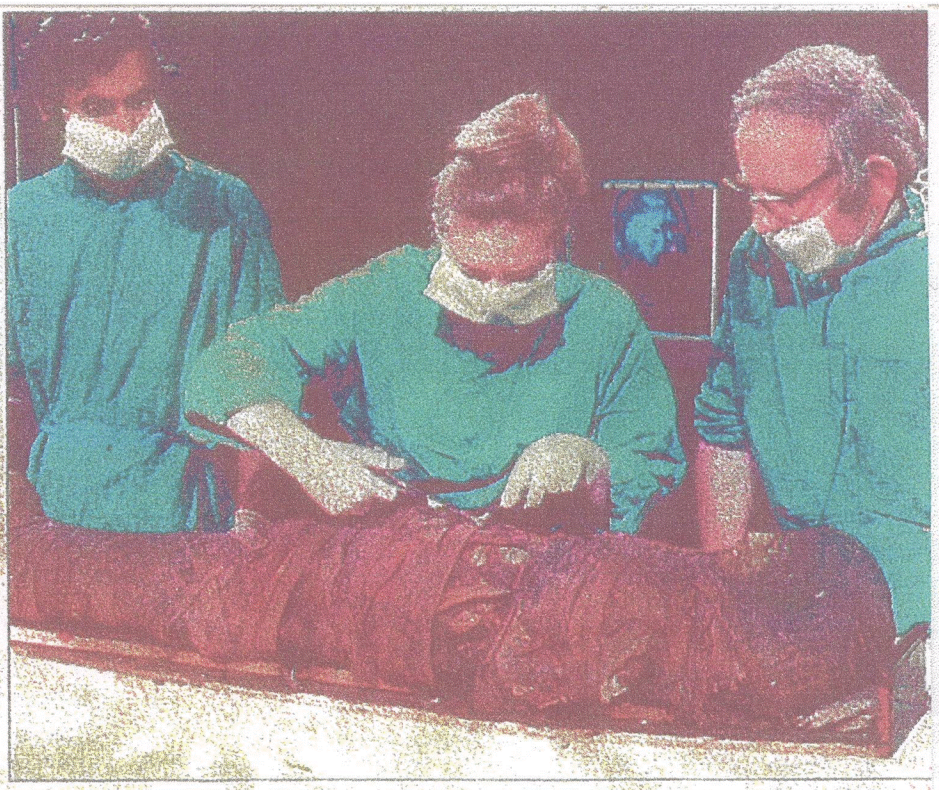


The study of genetics has revolutionised our understanding of human evolution. The older the specimen, the more we can learn about migrations of peoples, the spread of agriculture and of diseases, and even how we are all related. And now, says **Hilary Bower**, a new technique for extracting DNA means that even mummies are yielding some of their ancient secrets

MUMMY TO US ALL

MUMMIES DO NOT accommodate internal investigations very easily. Their innards are rigid and the scientist wielding the endoscope – a long a fibre-optic tube with a tiny video camera and scissors at its tip – must be highly skilled to coax it deep enough into the ancient body to allow a snip of tissue to be extracted. But coax they do. The Egyptologists from Manchester University have perfected this invasive but virtually non-destructive technique of retrieving precious samples of a human being long dead but alive with invisible information.

Eventually, says Dr Rosalie David and her team at Manchester University Museum, they hope to have a collection of some 8,000 endoscopic tissue samples – some more than 5,000 years old – from mummies all around the world, and a unique resource on which to practice the new investigative techniques that are transforming our knowledge of the past.

Based largely on genetics and immunology, these techniques promise to shed new light on everything from the spread of infectious disease and the development of agriculture to the origins and migrations of the human race itself. Dr David and her team are currently investigating DNA evidence of parasitical diseases such as schistosomiasis in the hope that tracking a disease's mutation over the centuries will reveal potential for new treatments.

"In ancient Egypt you got idealised artists' representations of people in the temples and monuments, because that's how they wanted to go on into the next life. But the reality of what their lives were like is right there in the bodies," she says. "In future, we'll be able to use genetic investigation to look at family relationships between mummies, to see how families were arranged and how movements of populations or foreign migration occurred at different times, either peacefully or by invasion."

There is also great potential for cross-cultural studies. "There are pre-Hispanic mummies in the Canary Islands and others in South America. There are bog bodies and frozen bodies, and naturally desiccated bodies in Mongolia which were found buried with tartan clothing and hats like the witches' hats of Europe. Instead of making educated guesses from literature, we'll be able to trace population movements quite widely."

According to Professor Robert Foley, head of the Human Evolutionary Biology Research Group in Cambridge, genetics has revolutionised the study of human evolution and history. "It is now possible to infer detailed aspects of human history from the distribution and frequency of genes found around the world today," he says. "Genes are like tracers, little markers on populations, and what we are trying to understand is the behaviours and the demographics and so on that have produced the patterns that they make in the genome."

The key process to illuminating these patterns is polymerase chain reaction (PCR), a lab test that works by amplifying tiny samples of DNA until there is enough available to read the genetic code and compare it with other samples. While medical geneticists search for gene mutations that link diseases and affected individuals, anthropologists, geographers and others look for "genetic variation" – simply the number of differences between a particular stretch of your DNA and mine, the DNA of someone on the other side of the world, or from someone who lived 100,000 years ago.

By counting such differences, it is possible to estimate how far groups of people are from a common ancestor, because mutations occur at a predictable rate. The more similar two individuals are, the more immediately they are related. The more different they are, the further back in time they shared an ancestor.

Tracing genetic variation is proving a valuable tool in some of anthropology's most fundamental questions. It has, for example, virtually settled the long-running and highly charged debate over whether modern humans evolved as a species in Africa, and then migrated out to replace other early hominids such as *Homo erectus*, or whether, in fact, we evolved from those early hominids.

"Undoubtably the best piece of work done on ancient genetic material was carried out last year, when researchers extracted and examined DNA from a Neanderthal bone to address the question of human origins," says Dr Mark Thomas, researcher and lecturer at University College London's Centre for Genetic Anthropology. "The big question was: were the Neanderthals – who were the European equivalent of *Homo erectus* and seem to have been in Europe at the same time as modern humans – within the human range of genetic variation, or were they outside and distinct? What the work on ancient DNA showed was that Neanderthals were in fact genetically distinct and that we are not the result of Neanderthals evolving into modern humans."

But it is not just ancient DNA that offers the potential to solve these puzzles from the past. Most genetic studies are currently being carried out on DNA collected from living people whose double helixes, experts say, hold clues to the entire history our race.

In 1986, scientists used mitochondrial DNA (see box opposite), which is only passed on through the female line, to calculate the date of what is described as the common ancestor of all human females – the "Mitochondrial Eve". More recently, a date has also been calculated for the "Y Chromosome Adam" using the genetic information that travels only through the male-line chromosome.

Although the dates are similar, says Dr Thomas, this does not mean that all humans sprouted from the loins of one single couple like the Biblical Adam and Eve. "Genetically, it's not required that they lived in the same place or at the same time. These are genetic realities, not human realities in the sense that we could think of them as common ancestors."

But the trail of mitochondrial or Y-chromosome analysis does allow discovery of past associations among far-flung populations – for if a particular type of either occurs in a number of different populations around the world, it's a sure bet that at some time those populations were in much closer contact.

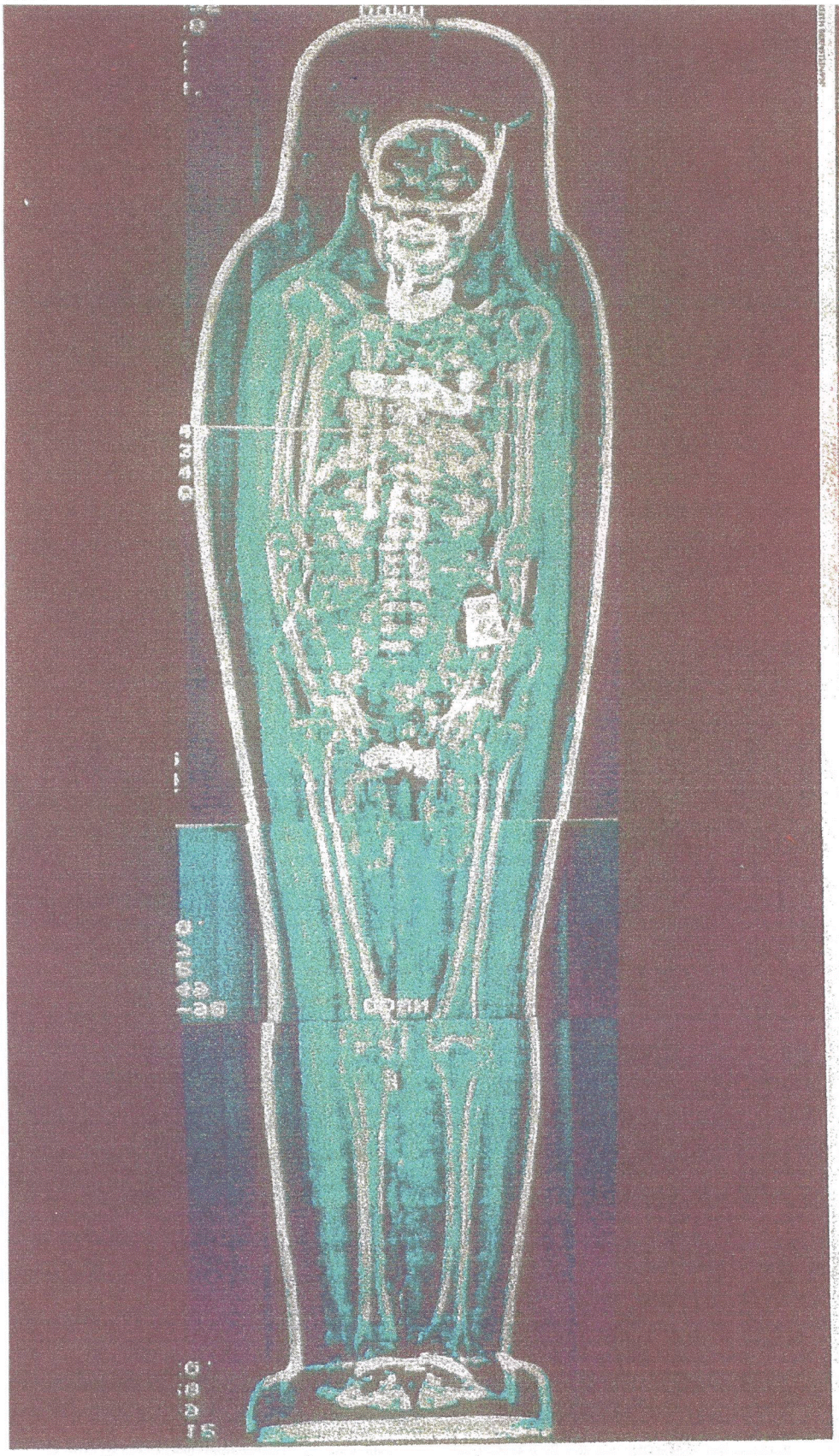
"One of the best examples of a nice genetic marker that always occurs within a population is found in the expansion of the Polynesians," says Dr Thomas. Thor Heyerdahl's Kon-Tiki expeditions popularised the notion that it was early South Americans who first peopled the Pacific, but DNA extracted from the bones of ancient Easter Islanders has been shown to contain a distinct genetic pattern that categorically links their origins to ancient Asia and not South America, Dr Thomas explains.

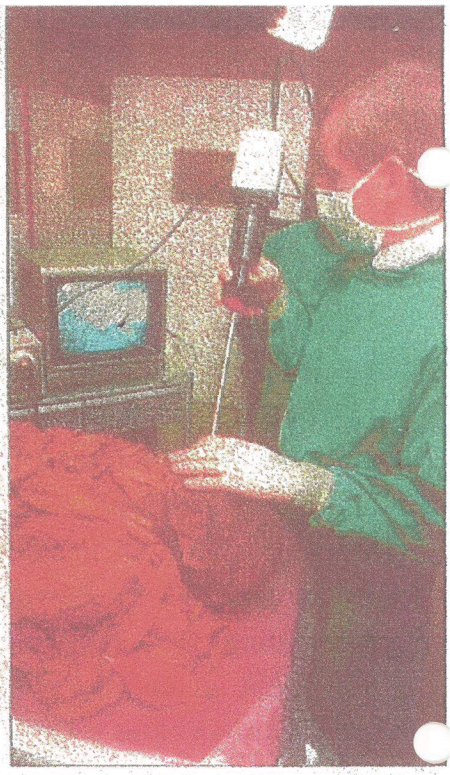
"From originating somewhere in South East Asia, probably around Taiwan and expanding as boat people all the way down through the Philippines to Melanesia, this group then rocketed their way straight into the Central Pacific – New Zealand, Hawaii, Easter Island. There has been a lot of archeological evidence for this expansion, but the genetic pattern really confirms it."

Another of anthropology's most cherished notions – that the Americas were first settled by people of Asian stock, who migrated across the Bering Strait land bridge, which linked what is now Siberia and Alaska during an ice-age fall in sea levels – is also under threat from new genetic knowledge.

Earlier this year, researchers from Emory University in Atlanta

Continued overleaf

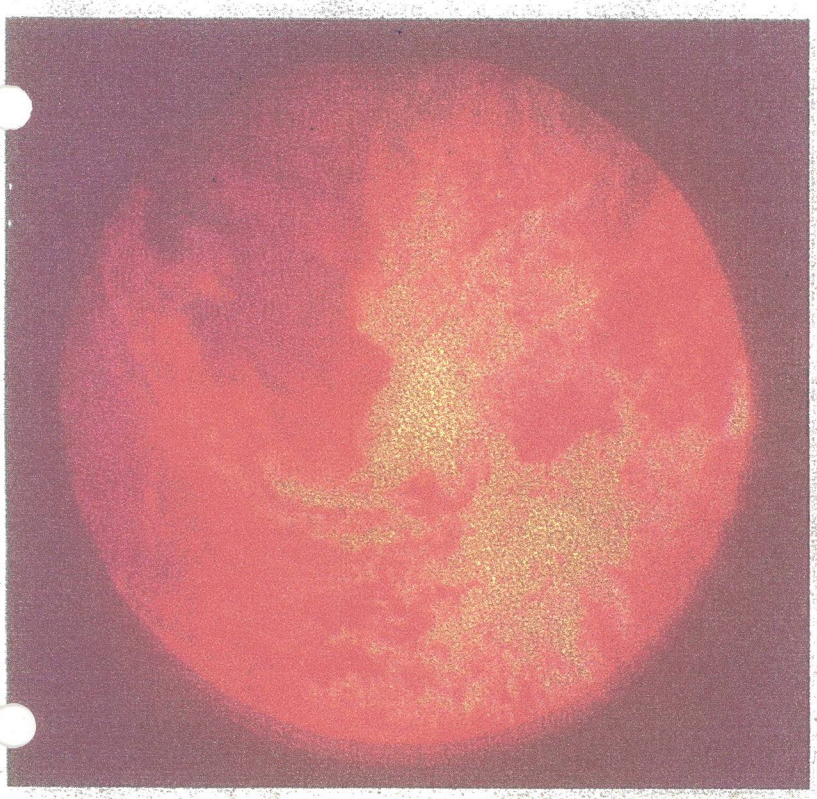




Yielding secrets: from left, Dr Rosalie David and members of the Manchester University Egyptology team unwrapping a mummy; an X-ray of a 3,000-year-old mummy; using an endoscope to obtain tissue samples, which removes the need to autopsy and destroy mummies

X +Y OF HISTORY

- Genes contain all the instructions needed to create a new individual.
- They are collected in 23 pairs of chromosomes. These are replicated in every cell in the body, except in sperm and egg cells, which carry only one member of each pair so that when the two come together in the act of fertilisation, the full set is restored.
- In 22 of the sets, the two chromosomes are so similar they can interchange bits between them. But the 23rd pair, called X and Y, are very different from each other.
- An egg always carries an X chromosome, but sperm carries either an X or a Y. An egg fertilised with an X sperm produces a female (XX); if fertilised with a Y sperm, a male (XY).
- A female's two X chromosomes can exchange bits between them, but a male's X and Y chromosomes cannot. This means the Y chromosome of each living man directly resembles that of his father, grandfather, great-grandfather and so on back into history, making analysis of the Y chromosome a powerful way to determine historical male lineages.
- In females, however, this doesn't work because interchange between the mother's and the father's X chromosomes muddies the genetic waters.
- But there is another part of our cells which is passed on solely through mothers. This is mitochondrial DNA: mitochondria are the microscopic "powerhouses" that provide a cell's energy and have their own separate DNA strand. Two children from the same mother will have identical mitochondrial DNA even if their fathers are different and so the female line can be traced right back to the "Mitochondrial Eve" (see main text).



The inside story: the chest area of a mummy, viewed through an endoscope, by Manchester University's team of Egyptologists

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reported that a genetic marker – labelled X and common in native Americans – is missing from the Asian genome, but is present in European groups, including Finns and Italians. It appears, say the researchers, that native Americans might be more closely related to the Europeans who desecrated their culture and stole their land than was ever imagined.

Genetic investigations can also be employed to check out the accuracy of historical dates indicated by literature or artefacts. In one study, Dr Thomas's team has been tracing the "Cohenim" – who were said to be the priests of the first Jewish Temple in Jerusalem and descendants of Moses' brother Aaron. "There are plenty of Cohens around today and in theory they should all have a common ancestry through their fathers going back to one person – Aaron.

"So we looked at the Y chromosome – which is only passed on through men – of Cohens from Jewish communities all around the world. What we found is that they are all much the same. And since part of the Y chromosome is known to mutate at a regular rate – at about 0.2 per cent per generation – we've been able to estimate the time distance from the common ancestor of all Cohens by looking at the number [illegible]anges there are between the diff-[illegible]t Cohens. We've now done this in Ashkenazi Cohens and in Sephardic Cohens and we got the same date." This biblical date, about 3,000 years ago, refers to the early temple period, Dr Thomas explains.

But it's not just population movements that can be traced by the study of genetic diversity. Population growth also leaves its mark on the genome, and is proving a fascinating tool for picking at such puzzles as how agriculture was first spread, and where certain diseases originated.

Piers Gollop, a PhD student at the Human Evolutionary Biology Research Group, explains: "Big populations hold an awful lot of genetic information. If you go to London and pick any two people at random, it's unlikely that they will share much similarity – in other words, be closely related. But if you go to a tiny village and select any two people at random, it's quite likely they will be related and will share genetic information. So if you compare sets of people from a global sample, where you see a big 'wave' or increase in the number of differences between them will tell you when the population expanded."

Such expansions can be linked to many things – climate, the spread of new knowledge, changes in land use, war, famine, says Gollop, who is studying how climactic changes such as glaciation and desertification in Africa might have influenced the development of different genetic strains now found in Europe.

Luigi Luca Cavalli-Sforza, the father of genetic anthropology, explains in his seminal book *The Great Human Diasporas* that the genetic markers of population explosions may also hold the key to the ongoing debate over how, where and when agriculture first developed, and whether it was spread by physical migration or by the transmission of ideas.

Cavalli-Sforza argues that genetic patterns support his theory: that agriculture so vastly increased the numbers of people that could be supported by a piece of land that local populations swiftly saturated the available resources and were forced to look elsewhere for more land. He believes that it was these migrants who took themselves and the practice of agriculture into Europe, eventually overwhelming the older hunter-gatherer species that were settled there. Others, however, argue that agricultural innovations were spread by travellers who returned home with new ideas, rather than via physical migration.

> IT APPEARS THAT NATIVE AMERICANS ARE MORE CLOSELY RELATED TO THE EUROPEANS WHO DESECRATED THEIR CULTURE AND STOLE THEIR LAND THAN EVER IMAGINED

Genetics is so useful in these fields, adds Cavalli-Sforza, because appearances and cultural similarities are unreliable indicators of genetic similarity. "Colonisers often convinced or coerced populations into taking up their cultures, but were themselves not numerous. Their contributions to the gene pool is limited and may not alter the genetic legacy of the peoples they encountered, for all their imposing visible legacy."

Relying on physical characteristics – face shapes, skin colour, body size and body shape – is risky too, since they are too easily modified by factors other than genetics, such as lifestyle and climactic changes.

Genetic information also aids the historical study of disease, according to Sonia Zakrzewski, whose work on disease in hunter-gatherer societies is supported by a Wellcome Trust bioarcheology grant. The spread of certain diseases is intimately linked to the development of agriculture, the increased contact with animals and the resulting sedentary lifestyle, she says.

"With genetic analysis, we can do a lot more tracking of the Neolithic Transition (when agriculture began). We know that's a time when a lot of diseases developed because people were in greater contact with animals and new plants. But many diseases don't affect skeletons so you can't detect them there, whereas you might be able to pick up traces of them in bones and soft tissue."

Tracking also makes it possible to date the origin of certain disease genes as well as those that are associated with resistance to certain diseases, casting light on the complex relationships between environment and genes – an obvious example being those which cause sickle-cell anaemia but also protect against malaria.

Health, disease, lifestyle, reproduction, migration: genetics, it appears, has infiltrated all the most intriguing questions of anthropology and geography, and its answers are likely to entertain scientists and spectators for some time to come. □