

From a Dry Bone to a Genetic Portrait: A Case Study of Sickle Cell Anemia

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ABSTRACT The potential and reliability of DNA analysis for the identification of human remains are demonstrated by the study of a recent bone sample, which represented a documented case of sickle cell anemia. β -globin gene sequences obtained from the specimen revealed homozygosity for the sickle cell mutation, proving the authenticity of the retrieved residual DNA. Further investigation of mitochondrial and Y chromosome DNA polymorphic markers indicated that this sample came from a male of maternal West African (possibly Yoruban) and paternal Bantu lineages. The medical record, which became available after the DNA analyses had been completed, revealed that it belonged to a Jamaican black male. These findings are consistent with this individual being a descendent of Africans brought to Jamaica during the trans-Atlantic slave trade. This study exemplifies how a "reverse population genetics" approach can be applied to reconstruct a genetic profile from a bone specimen of an unknown individual. *Am J Phys Anthropol* 111:153-163, 2000. © 2000 Wiley-Liss, Inc.

Developments of novel technologies in the recovery and analysis of DNA in human remains have contributed immensely to the field of molecular anthropology and forensic medicine. Mitochondrial DNA (mtDNA), HLA, autosomal, and later Y chromosome-specific mini- and microsatellite loci have been established as reliable tools for personal identification of missing people and victims of mass disasters, and in historical investigations (Hagelberg et al., 1991a; Jeffreys et al., 1992; Gill et al., 1994; Stoneking

et al., 1995; Cattaneo et al., 1997; Corach et al., 1997; Olaisen et al., 1997).

Despite these encouraging achievements, analysis of degraded DNA in forensic and archaeological specimens is still hampered by methodological difficulties and potential

The contribution of the first and the second authors is equal.

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pitfalls. The question of the authenticity of DNA isolated from human remains is the main concern. Trace amounts of highly damaged DNA in forensic and archaeological samples require the application of the extremely sensitive PCR method, which is prone to contamination. This problem may be overcome by applying a set of precautions and controls that have been worked out by the scientific community (Handt et al., 1994, 1996; Richards et al., 1995; Stoneking, 1995; Cooper, 1997).

We report here on the results of DNA analyses carried out on a bone specimen from an individual with documented (hemoglobin SS) sickle cell anemia, from the USA. The medical diagnosis was the only information available to the researchers prior to completion of the investigation. An interesting story unfolded in the course of the analyses of the β -globin gene, mtDNA sequences, Y chromosome DNA polymorphisms, and sex identification. The results provided a wealth of information about the individual, including his ethnic background and hereditary disease, which allowed us to draw a comprehensive genetic portrait. Corroborating evidence obtained from microscopic examination of the bone, as well as historical and medical records, facilitated a proper interpretation of the results and certified the authenticity of the genetic findings.

MATERIALS AND METHODS

Sample preparation

Two bone specimens from the left humerus and a thoracic vertebra were obtained during the autopsy¹ of an individual who had died during a sickle cell crisis in 1993. Following autopsy, flesh was removed from the bone specimens with a scalpel, which were boiled for 16 hr and then macerated in 10 times volume of 10% papain solution at 60°C overnight. The specimens were subsequently soaked for about 2 hr in warm dishwasher liquid solution, rinsed several times with warm water, and soaked for 2 hr in alcohol to facilitate air-drying. When dry, bones were soaked for 2 hr in ether to remove residual fat, and again

air-dried to allow the ether to evaporate. Finally, the specimens were placed in 3% hydrogen peroxide for 2 hr, rinsed for 3 hr in cool water, and air-dried.

Microscopic examination

Thin ground sections (50 and 70 μ m) were taken from the body of the thoracic vertebra (frontal section) and from the left humerus (transverse section) and prepared as previously described (Schultz, 1988). The sections were examined in plane and polarized transmission light, using a hilfsobject first order (quartz) as compensator.

DNA analyses

DNA extraction. Following histological examination, 0.5-cm-thick samples were removed from the thoracic vertebra (V) and humerus (H) under sterile conditions, immediately placed into plastic bags, and transported to the ancient DNA (aDNA) laboratory at the Hebrew University.

For each bone specimen, two separate DNA extracts were prepared and analyzed independently by at least two investigators several months apart. The DNA extracts obtained from the humerus were designated H-1 and H-2, and those from the vertebra were designated V-1 and V-2. The external layer of bones was removed with a sterile scalpel blade. Bone powder was obtained by drilling into the uncovered surface with a sterile bit. DNA was isolated from 1–3 mg of bone powder in 0.5 ml of freshly prepared DNA extraction solution, following the guanidinium-thiocyanate procedure (Höss and Pääbo, 1993).

Stringent precautions were undertaken to minimize possible contamination of the DNA extracts by exogenous DNA. These included physical separation of the work areas from those used for modern DNA analysis, and separation of the experimental steps (DNA extraction and PCR set-up were performed in two different daily ultraviolet (UV)-irradiated hoods, which were located in two different rooms separated from the analysis of PCR products and sequencing). In addition, only ultrapure reagents dedicated to aDNA analyses and sterile disposables and filtered tips were used while masks, gloves, and laboratory coats were worn. To monitor

¹Permission for unrestricted postmortem examination was granted by the patient's wife and legal next-of-kin.

possible contamination by nonrelevant DNA, appropriate controls were included for each step of the procedure: a "blank extraction" control containing no bone material, and a "blank PCR" control containing water instead of DNA extract. DNA samples of the investigators involved in the laboratory work were examined in parallel in a separate location in order to exclude false-positive findings resulting from handling.

PCR amplification and sequence analysis. Five microliters (if not indicated otherwise) of each DNA extract were subjected to hot-start PCR amplification (PCR Sprint Temperature Cycling System, Hybaid, Teddington, UK) in a 25- μ l reaction containing 20–50 ng of each primer, 0.2 mM each dNTP, 1.5–2.0 mM MgCl₂ (final), 0.8 mg/ml bovine serum albumin (BSA) (final), and 1 unit of AmpliTaq Gold DNA polymerase (Perkin Elmer, Oak Brook, IL). Amplification was performed for 50 (mtDNA)–55 cycles: 94°C for 30 sec, 50°C (for sex determination), and 53°C (for YAP⁺, sY81, and hypervariable segment I (HS I)) for 30 sec, and 72°C for 1 min.

A 179-bp fragment of the β -globin gene encompassing the complete first exon with the site of the A-T sickle cell transversion at codon 6 was amplified with the primers P11 5'-ACATTTGCTTCTGACACAACCT-3' and P14 5'-GGTCTCCTTAAACCTGTCTTG-3'.

The entire hypervariable segment I of the human mitochondrial control region was amplified in two overlapping fragments of 271- and 232-bp length, using the following sets of primers: set A, L 15985–5'-AGCAC-CCAAAGCTAAGATTC-3'-16004 and H 16255–5'-CTTTGGAGTTGCAGTTGATG-3'-16236; and set B, L 16191–5'-CCCATGCTTCAAGCAAGTA-3'-16210 and H 16422–5'-ATTGATTTACGGAGGATGG-3'-16403 (the latter as published by Horai et al., 1991). In two experiments, the complete HS I region was amplified in a single reaction with the primers L 16004 and H 16403, resulting in a 438-bp PCR product. The numbering refers to the Cambridge reference sequence (CRS) (Anderson et al., 1981).

Primers and PCR conditions for sex determination were as previously described (Faerman et al., 1995). The Y Alu polymorphism

(YAP) was analyzed with primers YAP1 and YAP2, yielding either a 150-bp PCR product for the YAP⁻ allele (without Alu insertion) or a ~450-bp fragment for the YAP⁺ allele (with Alu insertion) (Hammer and Horai, 1995). Moreover, the YAP⁺ allele was amplified as a ~230-bp fragment with primers AR237 and YAP2 (Hammer et al., 1997). The sY81 polymorphism was detected as described by Seielstad et al. (1994).

Eighteen-microliter aliquots of the PCR products were analyzed on 2% (for sex determination) or 1.5% NuSieve (FMC BioProducts, Rockland, ME) agarose gels, stained with ethidium bromide. Bands of the expected size were isolated from the gels with a sterile Pasteur pipette, and 3–5 μ l were used for direct ³³P terminator cycle sequencing (Amersham Life Science Inc., Arlington Heights, IL).

Typing of 12 Y chromosome DNA polymorphisms by multiplex PCR. Four DNA samples accompanied by the corresponding "no DNA extraction" controls were typed for 12 different Y chromosome DNA markers at University College London (UCL). The analysis was performed with two multiplex PCR kits designed for automated scanning of the amplification products (ABI-310TM genetic analyzer, Applied Biosystems, Foster City, CA) as described elsewhere (Thomas et al., 1998). In one kit, Y chromosome DNA variation was studied at the following six binary unique event polymorphisms (UEPs), including the two loci analyzed at the Hebrew University: YAP, sY81, Tat, 92r7, SRY+465, and SRY 4064. The microsatellite loci DYS19, DYS388, DYS390, DYS391, DYS392, and DYS393 were typed in the second kit. The PCR setup and conditions were as previously described (Thomas et al., 1998), except that the reaction volumes were 20 μ l, containing either 5 and 2.5 μ l (for the UEPs) or 1.3 μ l (for the microsatellites) of DNA extract and 0.1 mg/ml BSA. PCR was performed for 45 cycles.

RESULTS

Microscopic examination

The changes observed in both bone specimens were characteristic of chronic anemia

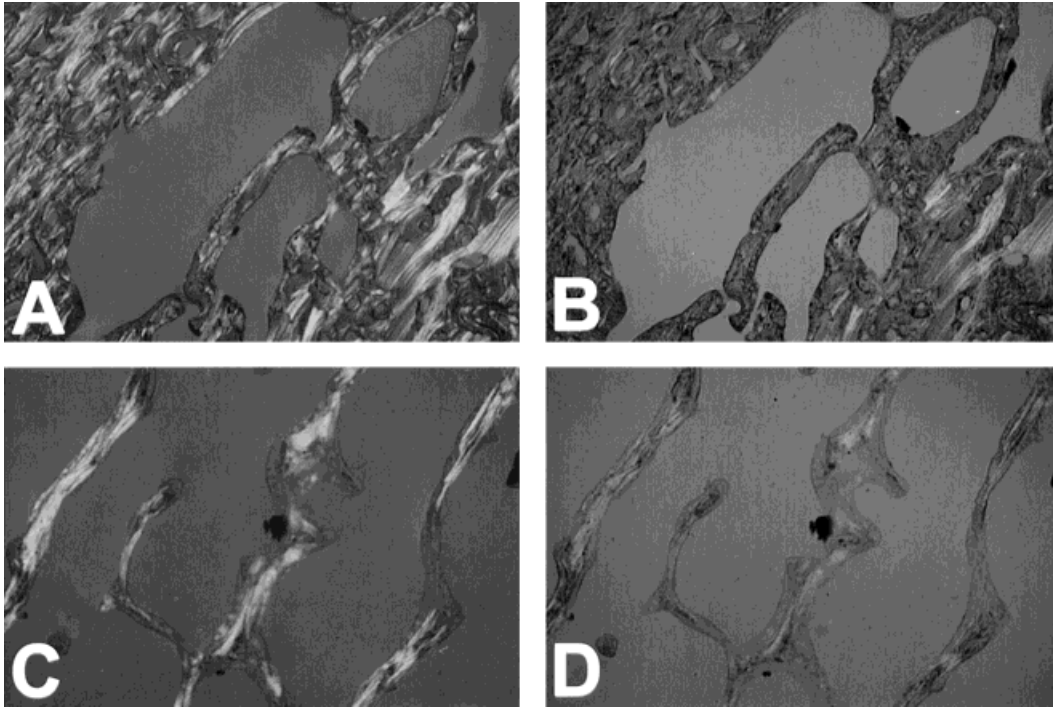


Fig. 1. Thin ground sections of the humerus (A, B) and of the vertebra (C, D) viewed in polarized transmission (left) and plane (right) light (magnification, $\times 25$).

(Resnick, 1981). The internal third of the left humerus, which should be relatively solid compact bone, was replaced by porotic areas, resembling spongy bone. The resultant cancellous-type modules (Fig. 1A,B) communicated with the diaphyseal medullary space. Thin ground sections microscopically examined under polarized light displayed orientations of collagen fibers. This was significant because it represented a record of successive clastic deletions and blastic apposition events. In the sample obtained from the humerus, the cancellous-type modules were clearly carved out from antecedent compact bone, presumably to afford additional space in which to house hyperplastic marrow. Furthermore, the contiguous diaphyseal medullary cavity contained coarse trabeculae in an area normally devoid of them. In the absence of any vestige of coarse fabric ("ischemic osteoid") here, some explanation other than bone infarct was sought. It was concluded that the trabecular had been retained since childhood, being formed at a time when this region of humerus was metaphyseal.

The sample taken from the vertebral body displayed an extremely thin cortical layer (Fig. 1C,D). Small portions of calcified cartilage were seen on the upper and the lower end plate of the vertebral body, as occurs in the normal aging process of discs. Bone trabeculae of the spongy structure were extraordinarily thin and relatively long, indicating atrophy. Consequently, cancellous modules of the vertebral marrow space were enlarged. Bony trabeculae in the mechanically more stressed area supporting the upper and lower end plates were comparatively dense, having a microstructure resembling a mosaic pattern, which is an indication of sustained, accelerated remodeling.

PCR and sequence analyses

We extracted DNA from two bone specimens: the cortical bone of the left humerus, and the spongy bone of the thoracic vertebral body. DNA extracts were then subjected to PCR amplification for the β -globin and amelogenin genes, the entire hypervariable segment I (HS I) of the human mitochon-

TABLE 1. Target sequences and size of PCR products¹

Target sequence	Size of PCR products, in bp	Size of PCR products, in bp			
		H-1	H-2	V-1	V-2
mtDNA, entire HS I	438	+	NT	NT	+
mtDNA, fragment B	271	+	+	NT	+
mtDNA, fragment A	232	+	+	NT	+
YAP ⁺ /YAP ⁻ allele ²	450/150	-/-	-/-	NT	-/-
X amelogenin allele	330	+	-	-	-
YAP ⁺ allele	230	+	NT	NT	+
Y amelogenin allele	218	+	+	+	+
sY81 ²	209	+	+	NT	+
DYS390 ³	200	+	NT	NT	+
β -globin gene	179	+	+	+	+

¹NT, amplification have not been performed.

²These loci were analyzed both at Hebrew University and UCL with identical results.

³Results for the largest microsatellite tested are presented here; for the rest, see Table 3.

drial control region, six UEPs, and six microsatellites loci. The DNA extracts yielded PCR products for all loci examined, despite harsh thermal and chemical preparation techniques (Table 1).

The 179-bp fragment of the β -globin gene was consistently obtained from all DNA extracts. The sickle cell A to T transversion at codon 6 was observed in the homozygous state both in the sense and antisense strands of the four PCR products (Fig. 2). Apart from the sickle cell mutation, no other changes in the β -globin sequence were noted. A known polymorphism at codon 2 found in the β -globin gene of one of the investigators was also not detected in the studied specimen. Since this polymorphism is not linked to any of the known sickle haplotypes and since none of the investigators carry the sickle mutation, these data together confirm the authenticity of the DNA analyses.

Amplification of the Y-amelogenin allele (218 bp) was successful in all four DNA samples (Fig. 3a). The extract H-1 also yielded the amplification product of the X (330 bp) allele (not shown). The individual was consistently identified as a male, while the analyses were carried out by females only.

DNA extracts H-1, H-2, and V-1 were used to amplify the fragments A (Fig. 3b) and B (not shown) that together spanned the entire HS I of the mitochondrial control region. The mtDNA fragments yielded a contiguous sequence between nucleotide positions 16008 and 16402, which showed the following nine substitutions relative to the CRS (Anderson

et al., 1981): C to G at position 16114, G to A at 16129, C to T at 16187, T to C at 16189, C to T at 16223, C to T at 16261, C to T at 16278, T to C at 16311, and C to T at 16360. When the extracts H-1 and V-2 were also tested for amplification of a larger, 438-bp product, both contained fragments of the expected size (not shown). Their sequences were identical to those obtained before.

Extracts H-1 and H-2 were first tested for the YAP and sY81 polymorphisms at Hebrew University. PCR with the primers YAP1 and YAP2, which amplify either the 150-bp YAP⁻ allele or the ~450-bp YAP⁺ allele, did not yield any product. In a reaction using the primers AR237 and YAP2, which detect a smaller fragment of the YAP⁺ allele (~230 bp), amplification products were obtained from both extracts (Fig. 3c). Sequence analysis of the two DNA fragments confirmed the presence of the YAP⁺ allele (Hammer, 1994). The poly(A) tail was found to contain 26 adenine residues, representing a YAP⁺ short allele. Aliquots of 10 μ l of H-1 and 8 μ l of V-2 DNA extracts gave amplification products for sY81. Both exhibited an A to G transition at position 168 (Fig. 4). Thus, the individual carried the haplotype YAP⁺ short and sY81-G, suggesting an African origin (Hammer et al., 1997).

For a more detailed Y chromosome haplotype, aliquots of extracts H-1, H-2, and V-2 with the corresponding extraction controls were coded and sent to UCL, where the analyses were performed blindly. Six UEPs, including YAP and sY81, and six microsatellites loci were typed. The status of each binary UEP marker scored after enzymatic digestion of the PCR products is given in Table 2. As expected, PCR products were not obtained for the YAP⁻ allele, the YAP⁺ allele was not detected because of its large size. The resulting haplotype was sY81-G, SRY4064-A, 92r7-C, Tat-T, SRY+465-C. The data for the microsatellite analyses are listed in Table 3. The microsatellite haplotype was 16-12-21-10-11-15.

Data analyses

The obtained sequence of the mtDNA control region was compared to those in several databases (GenBank, EMBL, and Concordance of Nucleotide Substitutions in the

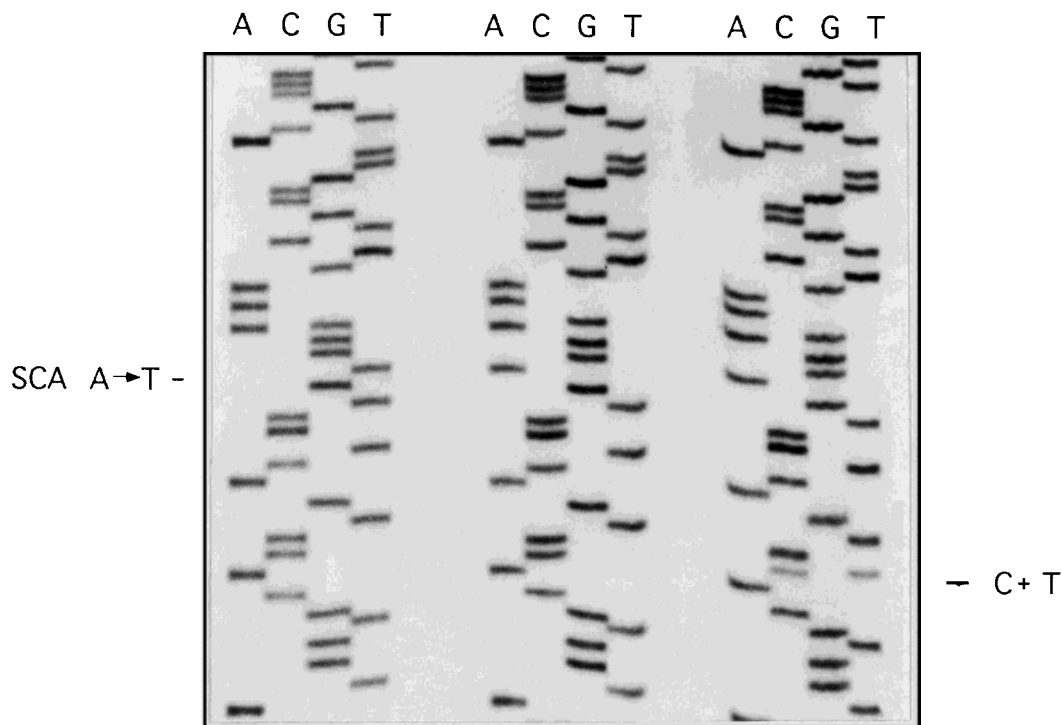


Fig. 2. Direct sequencing of the amplified β -globin gene fragment revealed the sickle cell A to T transversion at codon 6 in the homozygous state in the bone specimen (left) as compared to the normal (middle) sequence of investigator 1 and to that of investigator 2, showing a known C to C+T polymorphism at codon 2 (right).

Human mtDNA Control Region, University of Cambridge, UK). The sequence of the individual studied here was previously observed in an Afro-Caribbean (AFCR 58) and a Hispanic American (HISP 54, TWGDAM, quoted by Miller and Dawson, 1997). A single individual of the Yoruban population of present-day Nigeria (GenBank accession no. U94090) was found with an mtDNA haplotype different by a single substitution, lacking the C to G transversion at 16114 (Watson et al., 1996, 1997). This nucleotide position appears to mutate reasonably fast, as C to G/A transversions and, to a lesser extent, C to T transitions at 16114 have been observed in different mtDNA lineages in various ethnic groups (Di Rienzo and Wilson, 1991; Ward et al., 1991; Graven et al., 1995; Sajantila et al., 1995; Kolman et al., 1996; Richards et al., 1996; Watson et al., 1996). The Yoruban haplotype belongs to an African-specific cluster of diverse, phylogenetically

isolated mitochondrial lineages that are spread throughout Africa. These lineages are not shared between populations and are thought to represent relics of an ancient expansion event across the continent. The proposed most recent common ancestral type of this mtDNA cluster, dated to about 111,000 years ago, has a C at 16114 (Watson et al., 1997).

The mtDNA control sequence found in the present specimen can be considered to be derived from the ancestral type, without the transversion at 16114, that has so far only been reported in the Yoruban individual. Given Africa's large ethnic diversity and the paucity of comparable data on many of the different groups, it is at present very difficult to predict whether this particular haplotype is restricted solely to the Yoruban population, in which it was detected in 1 out of 33 tested individuals (Watson et al., 1997). In conjunction with historical accounts on the

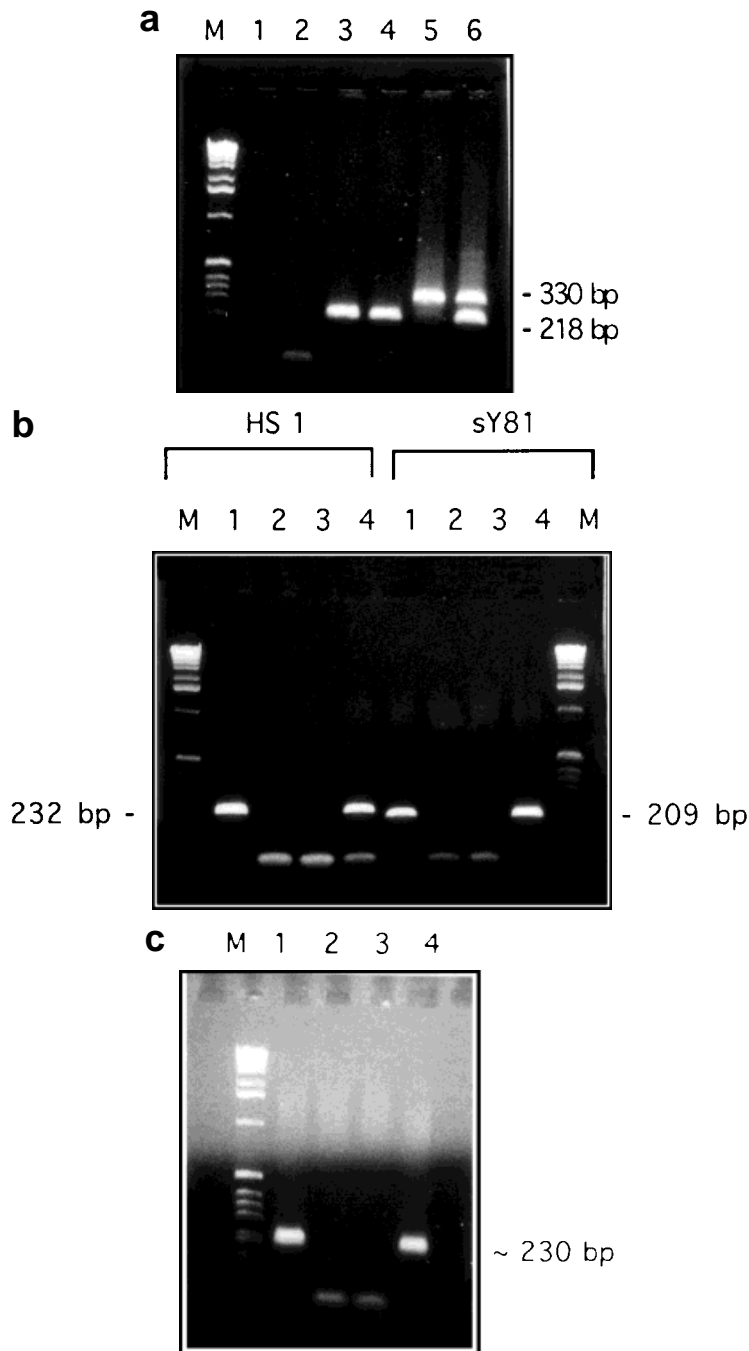


Fig. 3. PCR amplification of DNA recovered from the bone specimen. **a:** Sex identification. **Lane M**, 1-kb DNA size marker; **lane 1**, no DNA PCR control; **lane 2**, no DNA extraction control; **lanes 3, 4**, H-1 DNA sample; **lane 5**, modern female DNA control; **lane 6**, modern male DNA control. **b:** Amplification of the HS I region (left) and sY81 polymorphism (right). **Lane M**, 1-kb DNA size marker; **lane 1**, H-1 DNA sample; **lane 2**, no DNA PCR control; **lane 3**, no DNA extraction control; **lane 4**, modern DNA control. **c:** Amplification of the YAP⁺ allele. **Lane M**, 1-kb DNA size marker; **lane 1**, H-1 DNA sample; **lane 2**, no DNA PCR control; **lane 3**, no DNA extraction control; **lane 4**, modern DNA control.

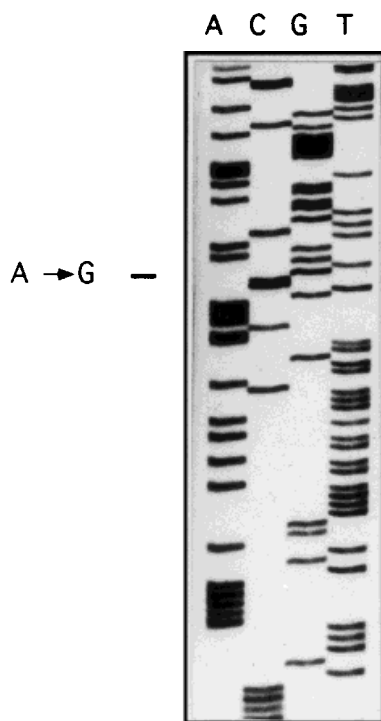


Fig. 4. Sequence analysis of the sY81 polymorphism in H-1 DNA sample, showing an A to G transition at position 168.

TABLE 2. UEP haplotype analysis¹

Extract	PCR	sY81	SRY4064	92r7	TAT	SRY+465
H-1	1	G	A	C	T	C
	2	G	A	C	T	C
H-2	1	NR	NR	NR	T	NR
	2	G	A	NR	T	C

¹NR, no reading.

slave trade, however, the presence of this mitochondrial lineage in our specimen and an Afro-Caribbean individual quoted by Miller and Dawson (1997) may very well point to a West African or even Yoruban maternal ancestry in the present case.

The Y chromosome UEP and microsatellite haplotype was compared to haplotypes in the Y chromosome database at UCL. The latter contains over 3,000 Y chromosome haplotypes from more than 40 populations from Africa, Asia, and Europe. Only four matches were found, three Bantu (Ndebele, Zulu, and North Sotho) and one Lemba, a Southern African tribe of enigmatic origin

known to have a substantial Bantu Y chromosome contribution to its gene pool (Spurdle and Jenkins, 1996). The compound Y chromosome microsatellite/biallelic marker haplotype is consistent with an African Bantu paternal ancestry of the individual.

Authenticity of DNA sequences

The authenticity of the findings is supported by several lines of evidence. We have never observed amplification products in numerous "blank extraction" or "no DNA" PCR controls set up in parallel to the specimen. Each of the results was consistent in two bone specimens from the same individual, and reproduced by at least two independent investigators several months apart. Moreover, the same typing was achieved for sY81 in two different laboratories. The observed microsatellite haplotype has only ever been found in YAP⁺ sY81-G individuals. Possible contamination of the specimen by laboratory personnel was ruled out on the basis of the β -globin, mtDNA, and sex identification results. We have never before analyzed in the aDNA laboratory a specimen with sickle cell anemia or a specimen of African origin. No Africans or African-Americans have entered or worked in the ancient DNA laboratory. Last but not least, we succeeded in confirming the medical diagnosis by identifying the sickle cell mutation in the β -globin gene.

It is generally agreed that the state of DNA preservation is not necessarily related to the age of the specimen (Hagelberg et al., 1991b; Faerman et al., 1995; Evison et al., 1997), that single-copy nuclear DNA survives less commonly than multicopy mtDNA (Cooper, 1997), and that with time, shorter fragments are more likely to be retrieved (Handt et al., 1996). Altogether, residual DNA recovered from this relatively recent bone sample demonstrated the same properties as DNA found in most forensic and archaeological specimens. Although amplification products were obtained for all loci examined, nuclear DNA fragments usually did not exceed ~220 bp, whereas twice-longer fragments of mtDNA were consistently amplified (Table 1). No amplification products were observed for the ~450-bp

TABLE 3. *Microsatellite haplotype analysis*¹

Extract	PCR	DYS19	DYS388	DYS390	DYS391	DYS392	DYS393
H-1	1	198.0 ²	126.2	200.3	163.1	164.6	126.8
	2	NR	126.2	200.3	163.5	164.6	126.9
V-2	1	197.8	126.1	200.1	163.2	164.6	126.8
	2	197.8	126.0	NR	163.3	164.6	126.7
Repeat number		16	12	21	10	11	15

¹NR, no reading.

²The sizes of the PCR products were obtained as GeneScan readings and converted into microsatellite repeat numbers according to Kayser et al. (1997), except for DYS388. For the latter, the repeat number was determined following Thomas et al. (1998).

YAP⁺ allele in the H-1 and V-2 DNA samples, although they yielded the 438-bp mtDNA sequence. The largest nuclear DNA product amplified was the 330-bp X amelogenin allele fragment obtained from the H-1 sample. PCR products as large as 179 and 218 bp were reproducibly amplified from all four DNA samples. We observed no differences in the amplification rate between the two bone specimens. In this specimen, DNA was equally well preserved in spongy and cortical areas in the bone samples.

DISCUSSION

Differential diagnosis of anemias in dry skeletal remains is difficult using traditional anthropological methodology. Analysis of β -globin sequences recovered from bone specimens with pathological lesions suggestive of anemia provides direct proof of the genetic mechanism causing the disease. This approach was successfully applied to identify a β -thalassemia mutation in an archaeological specimen with porotic hyperostosis, a condition traditionally attributed to severe anemia (Filon et al., 1995). In the present study, we have shown that the microscopic changes in the bone structure are consistent with the diagnosis of sickle cell disease, using DNA analysis. We suggest that microscopic examination, as part of preliminary screening, will facilitate the molecular detection of hemoglobinopathies in archaeological specimens with suspected anemia.

The mtDNA and Y chromosome haplotypes, obtained by means of ancient DNA analysis, permit inferences about the maternal and paternal origins of the individual. The results from the different markers complemented each other, pointing to an African ancestry. To summarize, the find-

ings indicate a male of maternal West African (possibly Yoruban) and paternal Bantu lineages, who was afflicted with sickle cell anemia.

Recent American history suggests that the individual might have been a descendent of Africans brought to the New World during the four centuries of the trans-Atlantic slave trade. Before abolition of slavery in the 19th century, kidnapped Africans came mostly from an area between present-day Senegal and Angola. The Yorubas of Nigeria in Central West Africa, one of the largest ethnic groups south of the Sahara, were especially affected by the slave trade (Bascom, 1969). For every decade from the 1760s onward, the Yorubas made up at least a fifth of all the slaves carried to the Americas (Curtin, 1969). Their proportion increased dramatically during the 19th century as a result of the Yoruban civil wars, when thousands of captives were sold into slavery. These persons were shipped mainly to the Caribbean Islands, and South and Central America, where Yoruban traditions survived and contributed immensely to the local cultures (Crowder, 1966; Bascom, 1969). Mitochondrial DNA sequence analysis comparing the Carib people of Belize with the Yorubas has indicated a considerable genetic input of these West Africans into the Native American gene pool (Monsalve and Hagelberg, 1997). This information prompted us to suggest that the studied individual might have had Caribbean roots. The assumption proved correct when the medical record, which had become available after completion of the DNA analyses, stated that the bone specimens belonged to a Jamaican black male who had lived in the USA prior to his death.

In his classic historical study, Curtin (1969) estimated that 68% of the 740,000 slaves imported into Jamaica between 1655–1807 had originated from Central West Africa, 17% from the Bantu-speaking areas and 15% from Atlantic West Africa. The percentages were calculated from shipping records listing the ports of origin in Africa. These data are in remarkably good agreement with those derived from the sickle cell haplotype analysis in present-day Jamaicans (Nagel and Ranney, 1990). Today, the highest frequency of sickle cell anemia worldwide is found in equatorial Africa, where the incidence of carriers reaches 35% (Bwibo and Kasili, 1982). The presence of the sickle trait in Jamaica is estimated at approximately 10% (Serjeant et al., 1986) and is considered to be the result of gene flow from Africa during the slave trade (Nagel and Ranney, 1990). It is worth mentioning that among the Yorubas of Nigeria, ~24% carry the sickle cell trait (Jelliffe and Humphreys, 1952).

Altogether, our findings indicating the presence of Central West African (Yoruban) and Bantu lineages in the Jamaican black individual are fully supported by several lines of historical evidence and agree very well with him being a descendent of Africans brought to Jamaica in the course of the trans-Atlantic slave trade. This study exemplifies how a “reverse population genetics” approach can be applied to reconstruct a genetic profile from a bone specimen of an unknown individual. This approach will be applicable to a wide range of questions in molecular anthropology as well as in forensics.

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