# Brief Communication: Y-Chromosome Haplotypes in Egypt

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*ABSTRACT* We analyzed Y-chromosome haplotypes in the Nile River Valley in Egypt in 274 unrelated males, using the p49a,f *Taq*I polymorphism. These individuals were born in three regions along the river: in Alexandria (the Delta and Lower Egypt), in Upper Egypt, and in Lower Nubia. Fifteen different p49a,f *Taq*I haplotypes are present in Egypt, the three most common being haplotype V (39.4%), haplotype XI (18.9%), and haplotype IV (13.9%). Haplotype V is a characteristic Arab haplotype, with a northern geographic distribution in Egypt in the Nile River Valley. Haplotype IV, characteristic of sub-Saharan populations, shows a southern geographic distribution in Egypt. Am J Phys Anthropol 121:63–66, 2003.


The Nile River Valley in Egypt offers several attractive features for studies in molecular anthropology. This region can be regarded as a long and narrow oasis, more than 1,000 km long, in which humans live distributed in a one-dimensional pattern, and where numerous population interactions can be expected to have taken place along the river. The comprehensive written record of Egyptian history (going back >5,000 years) affords an exceptional opportunity to compare spatial distributions of actual DNA markers with documented events. Moreover, the existence of large quantities of ancient skeletal and mummified remains offers, via ancient-DNA analysis, the potential for investigation of temporal variation.

The Nile River Valley had the potential to serve as a corridor for human migrations in Egypt, and there was a long-standing frontier zone between the northern and southern regions in Lower Nubia. Human populations distributed along the length of the Nile River Valley do exhibit cultural and linguistic differences. Both ancient Egyptian and Arabic are Afro-Asiatic languages, and Nubian languages constitute one branch of the Nilo-Saharan language family.

Recently, Krings et al. (1999) reported analysis of mitochondrial DNA (mtDNA) variation in contemporary Nile River Valley populations in Egypt, Nubia, and southern Sudan. The present study concerns a corresponding analysis of the paternal component, characterized by Y-chromosomal haplotypes, in Egypt.

## SUBJECTS AND MOLECULAR METHODS

Venous blood was obtained from a total of 274 unrelated adult males, living in Egypt during 1995–1999. The choice of these individuals was based on their locations in the valley, and in each case their genealogy goes back for several generations of paternally local ancestry. These 274 males included 162 inhabitants of Alexandria and the surrounding region (representating the Delta and Lower Egypt), 66 from Upper Egypt, and 46 from Lower Nubia (Fig. 1).

Genomic DNA was extracted from whole blood by the classic method, using proteinase K and several successive phenol-chloroform extractions (Gautreau et al., 1983).

We used the informative p49a,f Y-chromosome-specific DNA probes (Lucotte and Ngo, 1985), mapped to the nonrecombinant (NRY) Yq11.2 region (Quack et al., 1988). Using the Southern hybridization method, these probes oligolabeled by random priming revealed 10 male-specific *Taq*I fragments, at least five of which (*A, C, D, F,* and *I*) were polymorphic and determined 16 main haplotypes (I–XVI) in the initial reference population studied (Ngo et al., 1986).

## RESULTS

The frequencies of Y haplotypes in the three populations studied are given in Table 1. All 16 haplotypes are present in Egypt, except haplotype II. The main haplotype observed in Egypt is haplotype V, with a global frequency of 39.4% in the 274 samples

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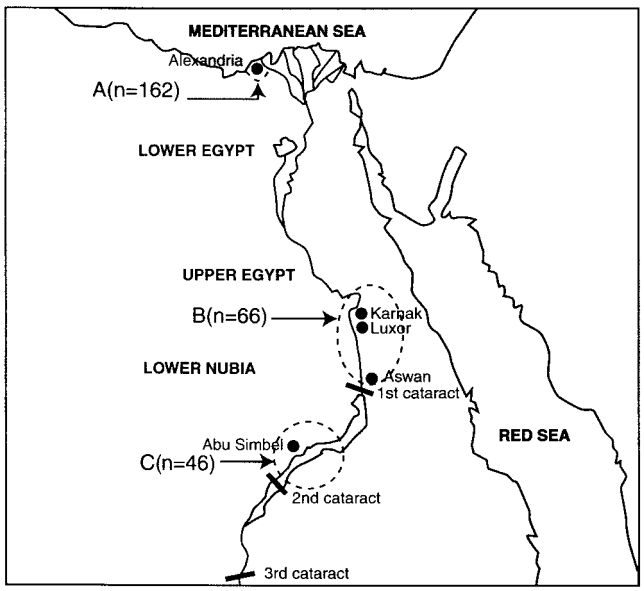


**Fig. 1.** Map of Egypt and Nubia, showing sampling localities along Nile River Valley. Actual political limit between Egypt and Sudan is across Aswan. Lower Nubia is considered to extend above that region. Sampling localities (with sample sizes n) are as follows: **A,** Alexandria (n = 162); **B,** Upper Egypt (n = 66); approximate territory of this group is indicated by an oval); **C,** Lower Nubia (n = 46), with territory as indicated by a circle (individuals originating from this locality self-identified as Nubians).

studied. The second haplotype in frequency is haplotype XI, with a global frequency of 18.9%. The third, fourth, and fifth haplotypes in frequency are haplotypes IV, VIII, and VII, with observed percentages of 13.9%, 7.3%, and 6.6%, respectively. Haplotype XV, with a global frequency of 5.5%, is observed in localities **A** and **B** only; haplotype XII, 2.2% in frequency, is observed in locality **A** only. Among the rarest haplotypes, haplotypes XIII and XIV are observed at 1.1% each, haplotypes III, VI, IX, X, and XVI are observed at 0.7% each, and haplotype I is observed only once, in locality **A**.

The geographic distribution of haplotypes (Table 2) is significantly heterogeneous ($\chi^2 = 25$, $df = 8$, $P < 0.001$) among the three localities. Chi-square analysis concerning comparisons of haplotype distributions among the localities shows that distribution of haplotypes in locality **A** is significantly different from haplotype distribution in locality **B** ($\chi^2 = 57$, $df = 4$, $P < 0.001$), and that haplotype distribution in locality **A** is significantly different from haplotype distribution in locality **C** ($\chi^2 = 77$, $df = 4$, $P < 0.001$); but haplotype distributions in localities **B** and **C** show nonsignificant differences between them ($\chi^2 = 2.5$).

Concerning comparisons between percentages of the 16 haplotypes in each of the three localities, the modal haplotypes are haplotype V in Alexandria (51.9%), haplotype XI in Upper Egypt (28.9%), and haplotype IV in Lower Nubia (39.1%).

The geographical distribution among localities is significantly heterogeneous between the three localities for haplotype V ($\chi^2 = 26.199$, $P < 0.001$), for haplotype XI ($\chi^2 = 13.595$, $P < 0.01$), and for haplotype IV ($\chi^2 = 56.149$, $P < 0.001$). Pairwise $\chi^2$ comparisons show that haplotype V is significantly more elevated in distribution ($P < 0.001$) in locality **A** compared to locality **B** (and also ($P < 0.001$), in locality **A** compared to locality **C**), but nonsignificantly different in distribution in locality **B** compared to locality **C**. The opposite is true for haplotype XI, which is significantly less elevated in distribution ($P < 0.01$) in locality **A** compared to locality **B** (and also in locality **A** compared to locality **C**, $P < 0.01$), and for haplotype IV, which is highly significantly less elevated in distribution ($P < 10^{-9}$) in both locality **A** compared to **B**, and **A** compared to **C**.

For haplotype VIII distribution, the $\chi^2$ test of heterogeneity is nonsignificant ($\chi^2 = 3.563$) between localities, as is also the case for haplotype VII ($\chi^2 = 3.022$) and haplotype XV ($\chi^2 = 1.326$). For haplotype XII, the $\chi^2$ value cannot be calculated because partial effectives are too low in value.

## DISCUSSION

Haplotypes V, XI, and IV are the main Y-chromosome-specific haplotypes in Egyptian males detected in the present study. Haplotype V is characteristic of Arab and Berber populations of North Africa (Lucotte et al., 2000), where it defines a major similarity among coastal populations in a one-dimensional pattern: the frequency of haplotype V is 53.4% in Tunisia, 56.7% in Algeria, and 57.9% in Morocco, reaching 68.9% among Moroccan Berbers where it is in the wide majority; the frequency of haplotype V is 44.7% in Libya, and was established to be 40.4% in a previously studied population of 52 males originating from the northern part of Egypt (Lucotte et al., 2000). Haplotype XI is one of the three most important haplotypes found in Ethiopia (Passarino et al., 1998; Lucotte and Smets, 1999), where it attains 25.9% in frequency. Haplotype IV is characteristic of sub-Saharan populations in Africa (Torroni et al., 1990; Spurdle and Jenkins, 1992), where its geographical distribution can be an indication of Bantu expansion: for example, in Central Africa (Lucotte et al., 1994), the frequency of haplotype IV is 55.2% in Cameroon, and reaches 80.3% in Zaïre and up to 83.9% in the Central African Republic.

In recent years, we assist to an explosion in human evolutionary studies driven by the many recently discovered polymorphisms on the NRY (Y Chromosome Consortium, 2002), since our initial description (Lucotte and Ngo, 1985) of the p49a,f *Taq*I restriction fragment length polymorphism's. Some of these studies on Y haplogroups (HG) or networks in Europe, based mainly on new polymerase chain reaction (PCR)-based binary polymorphisms, referred explicitly to our own original nomenclature system. For example, a clear analogy concerning the two main Y haplotypes in occidental Europe (Lell and Wallace, 2000) exists between HG1 and haplotype XV (Hill et al., 2000), and between

*TABLE 1. Y-chromosome haplotypes (classified in decreasing order of frequencies) observed among 274 Egyptians*

| Haplotypes | (Genotypes) | No. individuals observed in each locality | | | Total | (% obs.) |
|---|---|---|---|---|---|---|
| | | **A** | **B** | **C** | | |
| V | *(A2,C0,D0,F1, I1)* | 84 | 16 | 8 | 108 | (39.4) |
| XI | *(A3,C0,D0,F1, I1)* | 19 | 19 | 14 | 52 | (18.9) |
| IV | *(A1,C0,D0,F1, I1)* | 2 | 18 | 18 | 38 | (13.9) |
| VIII | *(A2,C0,D1,F1, I1)* | 17 | 2 | 1 | 20 | (7.3) |
| VII | *(A2,C0,D1,F1, I0)* | 14 | 3 | 1 | 18 | (6.6) |
| XV | *(A3,C1,D2,F1, I1)* | 11 | 4 | 0 | 15 | (5.5) |
| XII | *(A3,C0,D1,F1, I0)* | 6 | 0 | 0 | 6 | (2.2) |
| XIII | *(A3,C0,D1,F1, I1)* | 2 | 0 | 1 | 3 | (1.1) |
| XIV | *(A3,C1,D1,F1, I1)* | 2 | 1 | 0 | 3 | (1.1) |
| III | *(A1,C0,D0,F1, I0)* | 0 | 0 | 2 | 2 | (0.7) |
| VI | *(A2,C0,D1,F0, I1)* | 0 | 2 | 0 | 2 | (0.7) |
| IX | *(A2,C1,D0,F1, I1)* | 1 | 1 | 0 | 2 | (0.7) |
| X | *(A3,C0,D0,F1, I0)* | 2 | 0 | 0 | 2 | (0.7) |
| XVI | *(A4,C0,D1,F1, I0)* | 1 | 0 | 1 | 2 | (0.7) |
| I | *(A0,C0,D0,F1, I1)* | 1 | 0 | 0 | 1 | (0.4) |
| Total | | n = 162 | n = 66 | n = 46 | 274 | |

*TABLE 2. Distributions of haplotypes observed (and % expected) among three localities*[1]

| Haplotypes | No. individuals observed in each locality | | | Total (274) |
|---|---|---|---|---|
| | **A** | **B** | **C** | |
| V | 84 (63.9) | 16 (26.0) | 8 (18.1) | 108 |
| XI | 19 (30.7) | 19 (12.5) | 14 (8.7) | 52 |
| IV | 2 (22.5) | 18 (9.2) | 18 (6.4) | 38 |
| VII + VIII | 31 (22.5) | 5 (9.2) | 2 (6.4) | 38 |
| Others | 26 (22.5) | 8 (9.2) | 4 (6.4) | 38 |

[1] To obtain sufficient numbers of individuals, haplotypes VII and VIII were grouped together, as were haplotypes I, III, VI, IX–X, and XII–XVI, respectively.

HG3 and haplotype XI (Rosser et al., 2000); an analogy between HG21, or network 2.1 (Malaspina et al., 2000), and haplotype V is also evident. Haplotype VII is probably analogous to HG2, and haplotype VIII to HG9, but these haplogroups are more defined by what binary mutations they lack rather that by what mutations they have. Identity is absolute between network 1.3 (Malaspina et al., 2000) and haplotype XII.

As for mtDNA (Krings et al., 1999), the present study on the Y-chromosome haplotype shows that there are northern and southern Y-haplotypes in Egypt. The main Y-haplotype V is a northern haplotype, with a significantly different frequency in the north compared to the south of the country: frequencies of haplotype V are 51.9% in the Delta (location **A**), 24.2% in Upper Egypt (location **B**), and 17.4% in Lower Nubia (location **C**). On the other hand, haplotype IV is a typical southern haplotype, being almost absent in **A** (1.2%), and preponderant in **B** (27.3%) and **C** (39.1%). Haplotype XI also shows a preponderance in the south (in **C**, 30.4%; B, 28.8%) compared to the north (11.7% in **A**) of the country. In mtDNA, sequences of the first hypervariable *Hpa*I site at position 3592 allowed Krings et al. (1999) to designate each mtDNA as being of northern or southern affiliation, and proportions of northern and southern mtDNA differed significantly between Egypt, Nubia, and the Southern Sudan.

It is interesting to relate this peculiar north/south differentiation, a pattern of genetic variation deriving from the two uniparentally inherited genetic systems (mtDNA and Y chromosome), to specific historic events. Since the beginning of Egyptian history (3200–3100 B.C.), the legendary king Menes united Upper and Lower Egypt. Migration from north to south may coincide with the Pharaonic colonization of Nubia, which occurred initially during the Middle Kingdom (12th Dynasty, 1991–1785 B.C.), and more permanently during the New Kingdom, from the reign of Thotmosis III (1490–1437 B.C.). The main migration from south to north may coincide with the 25th Dynasty (730–655 B.C.), when kings from Napata (in Nubia) conquered Egypt.

Numerous postdynastic population influences, corresponding to additional migrations documented during the Ptolemic (300–200 B.C.)—Alexandria being a Macedonian city—Roman (since Egyptian annexation by Augustus), and later Arabic, Mameluk, and Ottoman times, are also likely to have contributed in a complex fashion to the current distribution of Y-chromosome haplotypes along the Nile River Valley.

Concerning less frequent Y-haplotypes in Egypt, haplotype VIII is characteristic of Semitic populations, originating in the Near East (Lucotte et al., 1993). For example (Lucotte et al., 1996), the frequency of haplotype VIII is 26.2% among North African Jews (where it represents the majority haplotype) and 77.5% among Jews from the island of Djerba (Tunisia), reaching 85.1% among Oriental (from Iraq, Iran, and Syria) Jews. Similarly, haplotype VII had a general geographical distribution fairly identical to that of haplotype VIII (which it often accompanies as a secondary haplotype); haplotype VII distinguishes itself by increased preponderance north of the Mediterranean and in Eastern Europe (Lucotte et al., 1996). Haplotype XV is the most widespread Y-haplotype in Western Europe (Lucotte and Hazout, 1996), where its frequency decreases from west to east (Semino et al., 1996; Lu-

cotte and Loirat, 1999). Haplotypes VIII, VII, and XV are less common haplotypes in Egypt (7.3%, 6.6%, and 5.5%, respectively), and tend to be located in the north of the country, near the Mediterranean coast. Possibly haplotypes VIII, VII, and XV represent, respectively, Near East, Greek, and Roman influences.

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