**Supplemental File S1. Materials and Methods**

*Population samples*

A total sample of 409 healthy individuals from Uganda participated in this work, under informed consent. During sampling, interviews were carried out in order to collect information concerning family relationships, and only maternally unrelated samples were selected. From the 409 samples, 109 were from Bantu and 300 from Nilo-Saharan speakers, the latter including 290 Nilotic speakers (243 Eastern and 47 Western Nilotic speakers) and 10 Central Sudanic speakers (Table S1).

All samples from Eastern Nilotic speakers, except for one individual, were collected in a particular region of East Uganda, Karamoja, characterized by an extremely arid climate able to support only a pastoralist lifestyle. They all belong to four ethnic groups: Karimojong (Bokora, Mazeniko and Pian), Jiye, Dodoth and Labwor. They were collected over a one-year period (2004-2005) from people attending the Moroto hospital, as well as during regular visits made by medical health care units to more than 50 different small villages spread over five of the seven districts of the Karamoja region (namely, Kaabong, Kotido, Abim, Moroto and Naikapiripirit). The communities sampled were typically composed of a small number of families and therefore just a few samples were collected in each village (4-10).A subset of these Eastern Nilotic samples (118 males) was previously studied for the Y chromosome ([Gomes et al. 2010](#_ENREF_27)).

A total of 19 samples from Western Nilotic speakers living in Karamoja were also collected in this region. 73 further Western (n = 7) and Eastern (n = 1) Nilotic, Central Sudanic (n = 2) and Bantu (n = 63) samples were collected in 2001-2003 from schools and colleges in the urban areas of the Kabale municipality surrounding Kabale’s largest market town in the Kigezi sub-region (estimated population size 41,350 according to the national population census of 2002) in the extreme southwest of Uganda. The remaining 75 samples were collected from Piida village in northwest Uganda, a village of population size 2361 at the time of collection in 1996, and included 46 Bantu (27 Mugungu, 16 Munyoro, 3 Nyarwanda) speakers, 8 Lendu Central Sudanic speakers and 21 Alur Luo Western Nilotic speakers, with ancestry in the West Nile region of northwest Uganda or the Democratic Republic of Congo.

*DNA extraction, PCR and sequencing analysis*

We extracted and analysed at IPATIMUP the 261 samples from Karamoja using a standard phenol-chloroform procedure, for the entire mtDNA control region (from positions 16024 to 16569; and from 1 to 576). We amplified this stretch in one reaction using the primers L15997 (5’–CCA CAT TAG CAC CCA AAG CT–3’) and H649 (5’–TTT GTT TAT GGG GTG ATG TGA–3’). The initial denaturation temperature was 95ºC for 15 min; followed by 35 cycles of 94ºC for 30s, 58ºC for 90s and 72ºC for 90s; with a final extension of 72ºC for 10 min. We performed the sequencing reaction using the Big-Dye Terminator v3.1 Cycle Sequencing kit (AB Applied Biosystems), and the following primers: L15997 (see above) and L16536 (5’–CCC ACA CGT TCC CCT TAA AT–3’) for forward sequencing reactions, and H649 (see above) and H036 (5’–CCC GTG AGT GGT TAA TAG GGT–3’) for reverse sequencing. After column purification with Sephadex™ G-50 (GE Healthcare) at 10%, we ran the sequenced products on an ABI PRISM 3130xl Genetic Analyser (AB Applied Biosystems) and analysed them with SeqScape software version 2.5 (AB Applied Biosystems) against the revised Cambridge reference sequence (rCRS) ([Andrews et al. 1999](#_ENREF_2)). We analysed the Kabale and Piida samples at the University of Huddersfield, for the HVS-I region (from positions 16011–16497; minimum extent of reads 16085–16430). We extracted DNA from buccal cells collected on sterile cytology brushes (Flowgen) using the InstaGene matrix (BioRad) according to the manufacturer’s protocol. We purified the PCR products using QIAquick PCR purification columns (Qiagen, Crawley, West Sussex) and sequenced them using a Beckman-Coulter CEQ8000 sequencer. We re-amplified purified PCR products using the relevant forward or reverse primer and Beckman-Coulter’s “Quickstart” master mix. We classified the mtDNA sequences into haplogroups following the most recent update of PhyloTree, mtDNA tree Build 16, 19 Feb 2014 ([van Oven and Kayser 2009](#_ENREF_61)), except where stated otherwise.

*Statistical analysis*

We compared the population samples from Uganda with African published data from the major African regions, as indicated in the table below.We restricted the calculation of genetic diversity indices and all other statistical analysis (including admixture, principal components, networks and search for shared haplotypes between populations) to the HVS-I segment, from positions 16090–16365, for which data were available for comparison with other African populations. We converted haplotypes found into sequences using the HaploSearch software (http://www.haplosite.com/haplosearch/) ([Fregel and Delgado 2011](#_ENREF_26)). We used the DnaSP 5.10 program ([Librado and Rozas 2009](#_ENREF_35)) to estimate diversity indices (except *θK*)*.* We calculated *θK* and pairwise *FST* genetic distances using the Arlequin 3.11 software ([Excoffier et al. 2005](#_ENREF_20)).

*Population samples for PCA*

For the PCA, we included Horn populations (Somalia, SOM; Ethiopia Afroasiatic: Omotic/Cushitic, ETA; Ethiopia unclassified, ETH); Eastern Nilotic speakers (Kenya: Maasai, ENM, Turkana, ENT; Ethiopia: Nyangatom, ENN); Kenya urban population (Nairobi, KUR); Western Nilotic speakers (Kenya: Luo WNL); Kenya Afroasiatic (Cushitic) speakers, KAA; Tanzania Afroasiatic (Cushitic) speakers, TZC; Tanzania Southern Nilotic speakers, TSN; Tanzania click-speakers (Hadza, TZH; Sandawe, TZS; Kenya Bantu speakers: Mijikenda, BKM and Taita, BKT (distinguished here as previously shown to be very distinct: Batai et al. 2013; Tanzania Bantu speakers, BTZ; Sudan/South Sudan (those of known provenance mostly the latter), SUD; East-Central African Bantu speakers (Rwanda: Hutu, BCE); East-Central African non-Bantu (DRC: Mbuti, PMB); Southeast Bantu speakers (Mozambique, BMZ; Zimbabwe: Shona, BZB; Zambia, BZM); Southwest Bantu speakers (Angola, BAN; Cabinda, BCB); West-Central Bantu speakers (Gabon North, BGN; Gabon West, BGW; Gabon Central, BGC; Gabon East, BGE; Gabon South, BGS; Cameroon BCM); West-Central Africa non-Bantu speakers (Chad, CHD; Ghana, GHA; Mali, MAL; Mali: Bambara, MAB; Mali: Malinke, MAM; Nigeria, NIG; Senegal, SEN; Sierra Leone, SIE; WC Tuareg, TUA; WC Fulani, WCF); North Africa (Egypt, EGY; Libya, LIB; Western Sahara/Mauritania, WSM; Morocco, MOR; Niger Fulani, NGF; Tunisia, TUN).

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Network code** | **PCA code** | **Region/Population Group** | ***n*** | **References** |
| **Uganda** | ENU | Eastern Nilotes | 243 | Present study |
| WNU | Western Nilotes | 47 | Present Study |
| BUG | Bantu | 109 | Present Study |
| **East-Central Africa Bantu speakers** | BRW  Description of the published African data used in diversity, PCA and Network analyses and corresponding population codes. | Rwanda/Hutu | 42 | Castri et al. ([2009](#_ENREF_10)) |
| **East Africa**  **Bantu speakers** | - | Kenya/Kikuyu | 22 | Watson et al. ([1997](#_ENREF_64)) |
| BKM | Kenya/Mijikenda | 192 | Batai et al. ([2013](#_ENREF_4)) |
| BKT | Kenya/Taita | 153 | Batai et al. ([2013](#_ENREF_4)) |
| BTZ | Tanzania | 58 | Knight et al. ([2003](#_ENREF_33)); Tishkoff et al. ([2007](#_ENREF_58)) |
| **Southeast Africa**  **Bantu speakers** | - | Madagascar | 40 | Tofanelli et al. ([2009](#_ENREF_59)) |
| BMZ | Mozambique | 408 | Pereira et al. ([2001](#_ENREF_41)); Salas et al. ([2002](#_ENREF_51)) |
| BZB | Zimbabwe/Shona | 59 | Castri et al. ([2009](#_ENREF_10)) |
| BZM | Zambia | 74 | de Filippo et al. ([2010](#_ENREF_18)) |
| **Southwest Africa**  **Bantu speakers** | BAN | Angola | 408 | Coelho et al. ([2009](#_ENREF_16)) |
| BCB | Cabinda (Angola) | 104 | Beleza et al. ([2005](#_ENREF_5)) |
| **West Central Africa**  **Bantu speakers** | BCM | Cameroon | 623 | Černý et al. ([2011](#_ENREF_13)); Quintana-Murci et al. ([2008](#_ENREF_47)) ; Veeramah et al. ([2010](#_ENREF_62)) |
| - | Equatorial Guinea/Bioko/Bubi | 43 | Mateu et al. ([1997](#_ENREF_37)) |
| BGC | Gabon (Central) | 135 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| BGE | Gabon (East) | 148 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| BGN | Gabon (North) | 113 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| BGS | Gabon (South) | 282 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| BGW | Gabon (West) | 156 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| - | Cameroon (Centre)/Baka | 127 | Quintana-Murci et al. ([2008](#_ENREF_47)) |
| - | Central African Republic/Biaka | 71 | Quintana-Murci et al. ([2008](#_ENREF_47)); Vigilant et al. ([1991](#_ENREF_63)) |
| - | Cameroon, Gabon/Bakola | 96 | Quintana-Murci et al. ([2008](#_ENREF_47)); Tishkoff et al. ([2007](#_ENREF_58)) |
| **East-Central Africa**  **Pygmy Nilo-Saharan speakers** | PMB | Democratic Republic of Congo/Mbuti | 51 | Quintana-Murci et al. ([2008](#_ENREF_47)); Vigilant et al. ([1991](#_ENREF_63)) |
| **Eastern Nilotic speakers** | ENM | Kenya/Maasai | 66 | Boattini et al. ([2013](#_ENREF_6)) |
| ENN | Ethiopia/Nyangatom | 89 | Poloni et al. ([2009](#_ENREF_45)) |
| - | Kenya/Samburu | 27 | Boattini et al. ([2013](#_ENREF_6)) |
| ENT | Kenya and Ethiopia/Turkana | 108 | Al-Abri et al. ([2012](#_ENREF_1)); Boattini et al. ([2013](#_ENREF_6)); Poloni et al. ([2009](#_ENREF_45)); Watson et al. ([1997](#_ENREF_64)) |
| **Western Nilotic speakers** | WNL | Kenya/Luo | 47 | Boattini et al. ([2013](#_ENREF_6)) |
| **Horn of Africa** | EDKETA | Ethiopia/Omotic and Cushitic | 115176 | Boattini et al. ([2013](#_ENREF_6)); Poloni et al. ([2009](#_ENREF_45)) |
| ETH | Ethiopia | 181 | Kivisild et al. ([2004](#_ENREF_32)); Soares et al. ([2012](#_ENREF_54)) |
| - | Ethiopia/Jew | 20 | Non et al. ([2011](#_ENREF_38)) |
| SOM | Somalia | 117 | Al-Abri et al. ([2012](#_ENREF_1)); Soares et al. ([2012](#_ENREF_54)); Watson et al. ([1997](#_ENREF_64)) |
| **East Africa non-Bantu or Nilotic speakers** | KAA | Kenya/Cushitic | 44 | Boattini et al. ([2013](#_ENREF_6)) |
| TZC | Tanzania/Cushitic | 47 | Knight et al. ([2003](#_ENREF_33)); Tishkoff et al. ([2007](#_ENREF_58)) |
| TZH | Tanzania/Hadza-Hadzabe | 117 | Knight et al. ([2003](#_ENREF_33)); Tishkoff et al. ([2007](#_ENREF_58)) |
| TZS | Tanzania/Sandawe | 80 | Tishkoff et al. ([2007](#_ENREF_58)) |
| KUR | Kenya/Urban | 92 | Brandstätter et al. ([2004](#_ENREF_8)) |
| **North Africa** | - | Algeria | 13 | Plaza et al. ([2003](#_ENREF_44)) |
| EGP | Egypt | 111 | Coudray et al. ([2009](#_ENREF_17)); Krings et al. ([1999](#_ENREF_34)); Saunier et al. ([2009](#_ENREF_53)); Stevanovitch et al. ([2004](#_ENREF_57)) |
| LIB | Libya | 73 | Fadhlaoui-Zid et al. ([2011](#_ENREF_22)) |
| WSM | Mauritania and Western Sahara | 66 | González et al. ([2006](#_ENREF_29)); Plaza et al. ([2003](#_ENREF_44)); Rando et al. ([1998](#_ENREF_48)) |
| MOR | Morocco | 223 | Brakez et al. ([2001](#_ENREF_7)); Coudray et al. ([2009](#_ENREF_17)); Falchi et al. ([2005](#_ENREF_23)); Harich et al. ([2010](#_ENREF_30)); Plaza et al. ([2003](#_ENREF_44)); Rando et al. ([1998](#_ENREF_48)); Rhouda et al. ([2009](#_ENREF_49)); Turchi et al. ([2009](#_ENREF_60)) |
| NGF | Niger/Fulani | 108 | Černý et al. ([2011](#_ENREF_13)) |
| - | Niger | 24 | Černý et al. ([2011](#_ENREF_13)) |
| NUB- | Egypt (Nubia) | 35 | Krings et al. ([1999](#_ENREF_34)) |
| TUA | Libya/Tuareg | 43 | Ottoni et al. ([2009](#_ENREF_39)) |
| TUN | Tunisia | 138 | Cherni et al. ([2009](#_ENREF_15)); Fadhlaoui-Zid et al. ([2004](#_ENREF_21)); Loueslati et al. ([2006](#_ENREF_36)); Plaza et al. ([2003](#_ENREF_44)); Turchi et al. ([2009](#_ENREF_60)) |
| **Southern Nilotic speakers** | SNT | Tanzania | 49 | Knight et al. ([2003](#_ENREF_33)); Tishkoff et al. ([2007](#_ENREF_58)) |
| **South Africa** | SKG- | South Africa/!Kung | 62 | Chen et al. ([2000](#_ENREF_14)); Watson et al. ([1997](#_ENREF_64)) |
| SKH- | South Africa/Khwe | 49 | Chen et al. ([2000](#_ENREF_14)); Tishkoff et al. ([2007](#_ENREF_58)) |
| **Sudan** | SUD | Sudan | 139 | Krings et al. ([1999](#_ENREF_34)); Soares et al. ([2012](#_ENREF_54)) |
| **West-Central Africa non-Bantu speakers** | CHD | Chad | 42 | Černý et al. ([2011](#_ENREF_13)) |
| GHA | Ghana | 227 | Veeramah et al. ([2010](#_ENREF_62)) |
| MAB | Mali/Bambara | 63 | Ely et al. ([2006](#_ENREF_19)); González et al. ([2006](#_ENREF_29)) |
| MAL | Mali | 57 | Černý et al. ([2011](#_ENREF_13)); González et al. ([2006](#_ENREF_29)) |
| MAM | Mali/Malinke | 86 | Ely et al. ([2006](#_ENREF_19)); González et al. ([2006](#_ENREF_29)) |
| NIG | Nigeria/Bula | 1293 | Černý et al. ([2011](#_ENREF_13)); Veeramah et al. ([2010](#_ENREF_62)) |
| SEN | Senegal | 148 | Rando et al. ([1998](#_ENREF_48)); Stefflova et al. ([2009](#_ENREF_56)) |
| SIE | Sierra Leone | 257 | Jackson et al. ([2005](#_ENREF_31)) |
| TUA | Burkina Faso, Mali and Republic of Niger/Tuareg | 42 | González et al. ([2006](#_ENREF_29)); Pereira et al. ([2010](#_ENREF_40)) |
| WCF | Burkina Faso, Cameroon, Chad and Mali/Fulani | 211 | Černý et al. ([2011](#_ENREF_13)) |

*Admixture analysis*

We carried out admixture analyses assuming that the mtDNA variation accumulated in the different Ugandan populations comes from one of the six main African regions: North, West-Central, Eastern, Southern, Southeast and Southwest Africa. Two different admixture models were considered: one using haplogroup frequencies and the other considering haplotype frequencies, as in Catelli et al. ([2011](#_ENREF_11)). We assumed the number of mtDNAs within each matching Ugandan haplogroup/haplotype (*ni*: 1 ≤ *i* ≤ *C*, the number of different matching haplogroups/haplotypes in the sample) to be a draw from a multinomial distribution with parameters and (1 ≤ *i* ≤ *C*), where *R* is the number of the source regions in Africa (North, West-Central, Southwest, Southeast, East, and South Africa), *fji* is the frequency of the *i*th haplogroup/haplotype in the *j*th source region (assumed to be known), and α*j* are the admixture coefficients ([see Salas et al. 2004](#_ENREF_52)). The probability of origin in each of the sub-continental regions can be computed as , where *n* is the number of Ugandan sequences with matches (≥ 1) in the whole database; *ki*, the number of times the sequence *i* is found in each Ugandan sample; *pis*, the frequency of the sequence *i* in each African region; and *pic*, the frequency of the sequence *i* in the whole database. We carried out the same analysis independently considering *n* to be the number of Ugandan sequences that have zero, one or two mutational differences from the sequences contained in the database; therefore, we considered *P0*, *P*1, and *P2* to be the admixture components when sequences matched perfectly, or differed by one mutational step, or two, respectively.

*Whole-mtDNA analysis*

We generated a total of 15 new L3i1 complete whole-mtDNA genome sequences (Table S3). Primer sequences, concentrations of each component in the PCR, thermo-cycling conditions, and analysis of mitogenomes were as previously described ([Cerezo et al. 2012](#_ENREF_12); [Gómez-Carballa et al. 2012](#_ENREF_28)). The new sequences are deposited in GenBank: KP229441–KP229455. Combining these with published sequences into a total data set of 26 whole L3i genomes, we reconstructed the phylogeny using the reduced-median algorithm ([Bandelt et al. 1995](#_ENREF_3)), with reticulations resolved by the relative mutation rates at each of position ([Soares et al. 2009](#_ENREF_55)). We estimated ages of clades using a time-dependent clock incorporating a correction for purifying selection ([Soares et al. 2009](#_ENREF_55)) using both the ρ statistic ([Forster et al. 1996](#_ENREF_25)) and maximum likelihood (ML) with PAML 3.13 ([Yang 1997](#_ENREF_65)), using a set of outgroups from L0a1b1, L1b1a, L2a1a, L3b1a, L3f1b, L3c, L3d1a, L3e1, L3k1, L3x1a2, L3h2 and L3a1 haplogroups (Table S4).

*Analysis of Ancestry Informative Markers (AIMs)*

In the present study we detected 10 samples belonging to non-(LxMN) mtDNA haplogroups, of which two are thought to have originated outside Africa (HV1b1 and T1a). To further investigate the overall genomic ancestry of these two samples, we tested them with a panel of 46 autosomal AIMs, previously developed to efficiently infer biogeographical ancestry and admixture proportions of four different continental origins (African, European, East Asian and Native American) ([Pereira et al. 2012](#_ENREF_42)), alongside control samples including four males with both confirmed African mtDNA and Y-chromosome haplogroups, plus two females with confirmed African mtDNA haplogroups.

We performed the genotyping of the 46 AIMs through multiplex PCR followed by capillary electrophoresis, as detailed in Pereira et al. ([2012](#_ENREF_42)). We then used the STRUCTURE v.2.3.3 software ([Falush et al. 2003](#_ENREF_24); [Pritchard et al. 2000](#_ENREF_46)) to perform clustering analysis and estimate membership proportions for the individuals carrying non-(LxMN) mtDNA haplogroups, while considering reference genotype data available for African, European, East Asian and Native American samples included in the HGDP–CEPH Diversity Panel subset H952 as the training set ([Cann et al. 2002](#_ENREF_9); [Pereira et al. 2012](#_ENREF_42); [Rosenberg 2006](#_ENREF_50)). Runs consisted of 100,000 Markov Chain steps after a burn-in length of 100,000 selecting the “use population information” option. We performed three independent replicates, assuming *K* = 4, which previous analyses suggested as the number of clusters that best fitted the reference population data (see Figure 2 in Pereira et al. 2012). In order to further evaluate the biogeographical ancestry assignment of the two questioned non-(LxMN) samples, we performed a PCA and calculated Bayesian likelihood values and likelihood ratios using the “Snipper 2.0 app suite” for the binary AIM classification of individuals (http://mathgene.usc.es/snipper/) ([Phillips et al. 2007](#_ENREF_43)).

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