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Title:

Population Genetics Histories of Central African Pygmies and non-Pygmies.¹

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“In reality, Pygmies do not exist. The humans that do exist are called Baka, BaBongo, BaKola, BaAka, BaSua, Efe, Asua, BaTwa... [...]”

Serge Bahuchet, “L’invention des Pygmées” (1993)

Anthropologists and ethnographers extensively described the cultural diversity of Central African Pygmy societies over the past 70 years. However, little is known about the origins of Pygmies and neighboring non-Pygmies mainly due to a lack of paleo-archaeological, historical and demographic data concerning the Congo Basin. In this chapter, we present how human population geneticists since the 1960’s have shed new lights on the previously unknown peopling history of Central Africa. In particular, we will show that interdisciplinary research between biologists and cultural anthropologists, rooted in joint field work, is a powerful tool to understand human biological diversity.

First, we introduce the main paradigms underlying population genetics studies of biological diversity and briefly illustrate several findings concerning human origins and migrations. Second, we highlight the lack of knowledge about the peopling history of Central Africa and describe the complex cultural criteria underlying the “Pygmy/non-Pygmy” population categorization in a multi-ethnic context. Third, we show how population genetics and ethnographic definitions of *a population* can be reconciled through interdisciplinary approaches and joint field work. Thus we establish the framework for studying Central African genetic diversity and use population genetics methods to reconstruct the peopling history of Pygmies and non-Pygmies. In particular, we show how population geneticists investigated questions regarding *i)* the common or independent origin of Central African Pygmy and non-Pygmy populations; *ii)* migration patterns among Central African populations; and *iii)* the evolution of differential adult height between Pygmies and non-Pygmies.

Population genetics approaches of human origins and past migrations

Where do I come from? Why do we all look so similar and yet so different? Most people may find these questions vertiginous and trying to address them is the fundamental challenge for human biologists. Since the 1950’s and the discovery of the chemical structure of DNA, one of

the major molecular basis of biologic inheritance, genetics provided rapidly growing sets of tools to reconstruct the history of mankind, especially when archaeological data are lacking. Any individual's DNA was inherited from half the DNA of each one of his/her parents, who themselves inherited their DNA from their respective parents, and so on at each generation. New mutations, i.e. new variants at a given location in the genome often called "genetic markers", arise randomly in the parental DNA and are transmitted to the offspring who will in turn transmit them to their descendants. An individual's DNA is thus a complex mosaic of DNA chunks inherited with mutations through the whole parental genealogy. This is why the DNA sequence of every existing individual is different² and why certain mutations are found in biological families and populations while absent in others. In a sense, the DNA sequence of an individual carries the coded history of his/her whole biological parenthood since the emergence of *Homo sapiens*. Therefore, by studying the distribution of given mutations across individuals, families, populations and groups of populations, population geneticists try to reconstruct the history of human lineages who gave birth to existing individuals. Finally, a small molecule of DNA is present in the mitochondria, a cell-compartment responsible, among other functions, for producing the energy necessary to the cell metabolism. This mitochondrial DNA is transmitted almost exclusively by mothers across generations. Therefore, by studying the mitochondrial DNA mutations, population geneticists focus specifically on the history of maternal lineages. In addition to the 22 pairs of DNA molecules (chromosomes) present in each cell nucleus of the organism, individuals carry a pair of sex-chromosomes. Males carry an X and a Y chromosome, the Y chromosome being transmitted exclusively from fathers to sons. Therefore, as opposed to mitochondrial DNA, mutations on the Y-chromosome carry the genealogical history of the whole paternal lineages.

The distribution of genetic variation across individuals is not only determined by how and when new mutations emerge or by the way parents transmit half of their DNA to their offspring. It is also influenced by natural selection forces. For an instance of positive natural selection, imagine a new mutation increasing the reproductive success of his/her carrier. This individual will have more children than individuals who do not carry this mutation and his/her children will in turn transmit this mutation to their numerous offspring. Generation after

² Except monozygotic twins who carry virtually the same DNA sequence.

generation, the frequency of this mutation will increase in the whole population. Conversely for negative natural selection, a mutation decreasing the reproductive success of individuals carrying them will decrease in population frequency over time. Therefore, population geneticists can study the distribution of genic mutations among existing individuals to identify potential signatures of past natural selection forces having influenced the evolution and adaptation of our species.

Demographic forces also strongly influence the distribution of mutations within and among populations. For instance, imagine that an individual dies accidentally before reproducing. If the population is of small census size, the unique mutations he/she carried may thus disappear from the whole population, whether these mutations provided a better reproductive success to the carrier or not. Conversely in a large population, several individuals are likely carrying some mutations inherited from their common ancestors. Therefore, the same “accidental” loss of individuals and mutations will affect the population frequency of mutations more slowly than in a small census population. This stochastic process of loss or fixation of mutations in a population, called genetic drift, thus strongly influences the evolution of population mutation frequencies over time, in interaction with population size, demographic expansions or contractions. Finally, migration and admixture between previously isolated populations are other major demographic forces influencing observed patterns of genetic diversity. For instance genetic variants private to a given population can appear in another population from where they were previously absent through the migration and mating of individuals. Therefore, by studying how neutral³ genetic variation is distributed across individuals among families and populations, population geneticists can infer the history of divergences, isolations, migrations, demographic expansions or contractions having likely given birth to existing human populations. Finally, by dating when mutations appear in individuals and genealogies, it is possible to date the occurrence of these various demographic events.

In this context, population genetics studies vastly furthered our understanding of human origins and biological evolution. Among the major findings of the past decades, it was shown

³ Genetic markers, or mutations, located far away from any known genes or functional genomic regions, a priori assumed to have evolved neutrally: i.e. without being direct targets of natural selection pressures but only influenced by demographic forces.

that the worldwide genetic diversity patterns were consistent with an African origin of all existing human populations. The current peopling of the world would result from an “Out of Africa” serial founder migration model over the past 100,000 years. In this model, mankind expanded throughout the world via series of migrations increasingly distant from the African source, each migration followed by founding events of new populations (Cavalli-Sforza and Feldman 2003, DeGiorgio et al. 2009). Interestingly, population geneticists found that an eastward migration initiated roughly 60,000 years ago via a coastal route out of the horn of Africa, in addition to an earlier migration route out of North-East Africa, could jointly explain the genetic patterns found in populations living nowadays in South-West, South-Central and South-East Asia (Macaulay et al. 2005, Quintana-Murci et al. 1999).

Importantly, population geneticists demonstrated that patterns of worldwide human genetic diversity strongly contradicted the existence of population-based biological races among humans. Indeed, the rapid colonization of the world from a common ancestral African population and numerous subsequent historical migrations and admixture between populations locally, have shaped the genetic diversity of mankind: the majority of human genetic variation (more than 95% on average across the genome) is found between individuals within populations rather than between individuals across populations (Cavalli-Sforza and Feldman 2003, Rosenberg et al. 2002). This shows that most of the phenotypic diversity observed between groups of populations, such as skin color, in fact grounds its genetic diversity basis within a very small proportion of the total genetic diversity found in mankind. In this context, the reader should constantly bear in mind that when population geneticists claim to have found “genetically differentiated populations”, this differentiation between individuals from different populations only stems from a very small proportion of the total genetic variation found in the whole sample.

The multiethnic context of Central Africa: Pygmy and non-Pygmy populations

African hunter-gatherer Pygmies are often seen as living examples of a life-style that dominated over 135,000 years of our evolution since the emergence of *Homo sapiens* roughly 150,000 years ago. Furthermore, Pygmies are currently neighboring populations with a widely different subsistence, agriculture, acquired much more recently in our evolutionary history, about

5,000 years ago in Central Africa. In this context, biological anthropologists have studied Central African populations over the past 50 years. In particular, they tried to evaluate the influence of life-styles and ecologies (e.g. parasitic or nutritional environments) on the biological and genetic diversity of human populations (Campbell and Tishkoff 2010, Froment 1993, Perry and Dominy 2009).

Archaeological records demonstrated that the Congo Basin has been densely and continuously inhabited by human populations since at least 45,000 years (Cornelissen 2002, Mercader 2003, Phillipson 2005). Nevertheless, how these past populations relate to the existing Pygmy and non-Pygmy populations remains largely unknown. This is essentially due to the lack of ancient human remains in this region, the acidic soil of the equatorial forest rapidly degrading bones. Therefore, biological anthropologists are challenged by a tremendous lack of historical and demographic knowledge, even recent, in Central Africa. Indeed, trying to understand how environmental pressures influenced the biological diversity observed across populations requires the prior knowledge of whether the studied populations share a common or independent origin and when the population divergence events occurred. For instance, knowing whether Pygmy populations have a common or independent origin is essential to understand whether biological features specific to several Pygmy populations evolved recently in each population separately or whether these features were inherited from a common ancient ancestral population.

The binary categorization of Central African populations into Pygmies and non-Pygmies is deeply rooted in the Western culture. The ancient Greek poet Homer first introduced the word Pygmy to refer to an anthropomorphic mythical population in the epic narrative Iliad (Song 3 v. 1-6). He briefly describes the Pygmies as a population of short stature individuals (the word Pygmy refers to a length or volume measurement such as a cubit or a handful), living somewhere overseas, and at war with migrating cranes. Western explorers travelling across Central Africa during the second half of the 19th century encountered numerous short stature populations and designated them as Pygmies in a scholarly reference to Homer (Bahuchet 1993, Du Chaillu and Owen 1867, Schweinfurth 1873).

During the second half of the 20th century, anthropologists described *a posteriori* the numerous and complex cultural criteria underlying the historical Pygmy/non-Pygmy categorization of Central African populations (see other chapters in this volume and Bahuchet

1993, Bahuchet 2012, Froment 1993, Hewlett 1996, Hewlett et al. 1982, Joiris 2003). They showed that populations could be designated as Pygmies distinctly from neighboring non-Pygmies if: *i*) they designate themselves with a different ethno-name than neighboring ethnic groups; *ii*) they are designated as a different ethnic group by neighbors; *iii*) they are recognized by outsiders as specialists of forest activities, such as hunting, gathering and fishing, and possess an extensive knowledge of forest environments (both ecological and esoterical); *iv*) they are very mobile in their environment; *v*) they maintain complex socioeconomic relationships with specific neighbors, including the trading of goods, exchanges of workforce and sometimes participation in social and ritual events; *vi*) they are recognized as outstanding musicians sometimes with specific musical instruments and vocal techniques. Additionally, physical anthropologists demonstrated that populations categorized as Pygmies based on these cultural criteria were among the shortest worldwide, despite a tremendous variability in average stature across Pygmy populations (Froment 1993, Hiernaux 1974).

Most importantly, ethnographers and anthropologists extensively demonstrated that cultural features were highly variable across Pygmy populations. Therefore, the Pygmy/non-Pygmy categorization is highly reductive of an often much more complex reality. For instance, the various groups historically designated as Pygmies do not share a common language today. In fact, they all speak different languages including languages from different linguistic families (Bahuchet 2012). Furthermore, Pygmies do not have a common myth of origin and rarely know of other populations also called Pygmies by outsiders. The wide cultural and morphological diversity across Pygmy populations *a priori* challenges the common view that all Pygmy populations throughout Central Africa share a common heritage or origin, a view *de facto* assumed by the widespread use of the umbrella term “Pygmy”.

In summary, the peopling history of Central Africa and the origins of Pygmy and non-Pygmy populations remain widely unknown. This is mainly due to the lack of historical and archaeological data as well as the complex cultural features underlying the Pygmy/non-Pygmy categories in the multiethnic context of Central Africa. In this context, population genetics approaches provide a promising set of methods and tools to try to reconstruct indirectly the past origins of existing Central African Pygmy and non-Pygmy populations.

Reconciling the definitions of *a population* between research fields

We have previously shown that population genetics methods can be used to infer the past demographic history of populations by studying patterns of genetic diversity within and among existing populations. Similarly as in cultural anthropology disciplines, all the observations and conclusions drawn by geneticists about the diversity and origins of human groups rely on how a population is defined. Population geneticists, since they are studying the history of the genes carried by existing individuals (their genealogies literally), intuitively define a population as a group of individuals more likely to mate with each other than to mate with other groups, albeit not necessarily strictly.

The multiethnic context of Central Africa exemplifies how the complex and dynamic cultural-based definition of ethnic groups and populations can be at odds with this genetic-based definition. As previously described, numerous culturally heterogeneous and geographically isolated populations are gathered under the term Pygmy. Merging *a priori* all DNA samples obtained from various Pygmy populations in a single population labeled “Pygmies” can thus be strongly misleading for population genetics studies since the individuals here merged may belong to distinct, mutually exclusive, reproductive units or population genetics “populations”. In order to apply population genetics methods in Central Africa it is therefore necessary to precisely assess how the DNA samples collected from individuals relate to *i*) existing populations as defined *a priori* in population genetics approaches and *ii*) the complex Pygmy/non-Pygmy cultural categorization established by anthropologists and ethnographers.

This can be achieved by interdisciplinary field work between ethnographers and population geneticists and sampling jointly DNA and ethnographic data such as ethnicity, family structures, marriage customs and mobility of spouses, life-style, etc. Familial anthropology data as well as knowledge of marriage customs and the mobility of spouses allow geneticists to identify *a priori* groups of sampled individuals more likely to mate with each other than they are to mate with other communities. Furthermore, detailed ethnographic data on cultural features such as endogenous and exogenous ethnic representations, life-styles or socioeconomic interactions between neighboring communities, further allow population geneticists to accurately categorize sampled individuals and populations in Pygmies or non-Pygmies based on ethnographic criteria. Such interdisciplinary approach was first developed to study Central

African Pygmies and non-Pygmies by Hewlett and Cavalli-Sforza starting in the 1960 - 1970's. These authors established a powerful framework to study Central African genetic diversity. It further allowed studying the complex relationships between a variety of cultural and biological phenomenon such as how the widespread cultural barriers against marriages between Pygmies and non-Pygmies translate (or not) into genetic isolation between neighboring communities.

First, we present what population genetics approaches rooted in interdisciplinary field work revealed about the origins and biological diversity of Central African Pygmy and non-Pygmy populations. Despite ethnographers and anthropologists repeated efforts, the oversimplistic myth of the short-stature nomadic forest hunter-gatherer Pygmies isolated from the rest of the world still overwhelms the imagination of Westerners⁴. We then present how population geneticists faced these myths to study patterns of genetic admixture among Central African populations and the differential biological evolution and adaptation of Pygmies and non-Pygmies through the example of height.

I. The genetic diversity and origins of Pygmies

Starting in the late 60's, Cavalli-Sforza and collaborators were the first population geneticists to systematically study the genetic diversity of several Central African Pygmy populations. Using a reduced number of genetic markers underlying the various blood groups present in Western (CAR and Cameroon) and Eastern (Western Democratic Republic of Congo) Pygmy groups, these authors first found that Pygmy populations were highly differentiated from other Central African non-Pygmy populations, although Pygmies indisputably exhibited a genetic pattern of African origin. Second, these authors identified major genetic differences between the various Eastern and Western Pygmy populations as well as among Western Pygmy groups (Cavalli-Sforza et al. 1969, Santachiara-Benerecetti et al. 1980). Nevertheless, all these groups shared specific blood-types and other protein-related genetic patterns that led Cavalli-Sforza and colleagues to conclude in favor of a likely ancient common origin of all Pygmy populations, followed by a more recent split between Eastern and Western Pygmy populations.

⁴ illustrated for instance among many others by the first Tarzan movie: *Tarzan the Ape Man* (dir. W. S. Van Dyke, 1932). Further see chapters X and X in this book.

In this model, the differential influence of genetic drift after the East-West split and recent admixture between Pygmies and neighboring non-Pygmies explained the genetic differentiation found among Pygmies today (Cavalli-Sforza 1986, Cavalli-Sforza et al. 1977, Cavalli-Sforza et al. 1969, Daiger and Cavalli-Sforza 1977, Santachiara-Benerecetti et al. 1980). Cavalli-Sforza and colleagues could not test formally these hypotheses or obtain precise estimate for the divergence times and admixture proportions due to the limited number of genetic markers and the lack of statistical and methodological tools available at the time. Nevertheless, these studies posed the initial questions for all further population genetics studies of Central Africa: when did Pygmy populations diverge from non-Pygmy populations? When did Eastern and Western Pygmy populations split? When and how Western Pygmy populations started their genetic differentiation? Are Pygmy populations genetically isolated from neighboring non-Pygmies?

The Aka (CAR) and Mbuti (Eastern DRC) Pygmy DNA samples collected by Hewlett and Cavalli-Sforza in the 1980's during interdisciplinary field work were later included in wider worldwide genetic databases⁵. They were studied alongside other African and non-African populations in the emerging systematic investigation of human worldwide genetic diversity (Cann et al. 2002, Cavalli-Sforza 2005). Despite the growing number of studies showing that these two Pygmy populations exhibited largely different genetic patterns, a very limited number of studies tried to tackle the question of the origins of Pygmies until the 21st century, perhaps due to the belief that this question had already been definitively solved by the extensive work of Cavalli-Sforza and colleagues.

Giovani Destro-Bisol and colleagues were among the first to re-discover the issue of the origins of Pygmies, with a series of articles published between 2000 and 2004, and tried to precisely characterize the genetic diversity among the available population samples (Destro-Bisol et al. 2000, Destro-Bisol et al. 2004a, Destro-Bisol et al. 2004b). They found that the three Pygmy populations under study (the Aka and Mbuti samples previously gathered by Cavalli-Sforza and Hewlett and a third sample of Aka Mbenzele Pygmies from South-Western CAR) had widely differentiated mitochondrial DNA, but that they shared a number of markers indicating an ancient common maternal origin (Destro-Bisol et al. 2004a). Assuming a common origin of

⁵ The Human Genome Diversity Project - Centre d'Etude du Polymorphisme Humain (HGDP-CEPH) encompasses more than a thousand individual DNA samples from more than 50 populations across Africa, Middle-East, Europe, Asia, Oceania and the Americas.

Eastern and Western Pygmy populations, the authors simulated genetic data and found that the ancestral maternal lineage split roughly 70,000 years ago into two lineages respectively giving birth to Pygmy and non-Pygmy populations. They further estimated that Eastern and Western Pygmy groups of populations diverged from the ancestral Pygmy population between 3,000 and 18,000 years ago. However, these studies did not fundamentally question the initial hypothesis of a common origin of Pygmies. In fact, the lack of both population samples and genome wide genetic markers made it difficult to test this hypothesis.

a. Genetic differentiation among Central African Pygmy populations

To overcome these limitations, numerous interdisciplinary fieldwork, following Hewlett and Cavalli-Sforza initial approach, were conducted mainly by French and American researchers. Between 2002 and 2009, researchers sampled DNA from more than 20 populations in Western and Eastern Central Africa (Batini et al. 2011, Patin et al. 2009, Tishkoff et al. 2009, Verdu et al. 2009, see Figure 1 and Table 1). These populations were categorized as Pygmies and non-Pygmies based on the numerous cultural criteria set by ethnographers and anthropologists.

Considering 26 Central African populations (728 individuals, Table 1 and Figure 1), we study here the genetic diversity patterns using 26 independent neutral markers genome-wide (see Table 1 and Figure 2 legend). We found that certain Pygmy and non-Pygmy pairs of populations exhibit the largest genetic differentiation levels in our data set even when considering only Cameroonian and Gabonese populations (Figure 2). Furthermore, most of the genetic variation explained by differences among populations (1.66% of the total genetic variation AMOVA F_{ST} , Excoffier et al. 1992), was accounted for by the major differentiation among Pygmy populations (2.25% of the total genetic variation when considering only 13 Pygmy populations). Conversely, non-Pygmy populations exhibited small, often non-significant levels of population genetic differentiation (0.46% of the total genetic variation when considering only 13 non-Pygmy populations). Finally, Eastern and Western Pygmy groups of Pygmy populations exhibit major genetic differentiation, followed by high levels of between population differentiation respectively among Western and Eastern Pygmy groups (see Figure 2 and Patin et al. 2009, Tishkoff et al. 2009, Verdu et al. 2009). These results significantly expanded previous observations by Cavalli-Sforza, Destro-Bisol and collaborators and echo the vast cultural diversity found by ethnographers among populations designated as Pygmies. Furthermore, while

some Pygmy populations showed large genetic distances with non-Pygmy populations, some others, such as the two Gabonese Bongo populations, were found resembling genetically non-Pygmy neighbors (Figure 2 and Verdu et al. 2009). These results further jeopardized the traditional view that all Pygmies are biologically homogeneous and widely different from their neighbors.

b. Common genetic origins of Central African Pygmies

In this context, we formally tested for the first time whether genetic patterns observed among numerous Pygmy populations from Cameroon and Gabon were consistent with a common origin of Pygmy populations or with a historical scenario where each existing Pygmy population diverged independently from the lineage that gave birth to non-Pygmy populations (Verdu et al. 2009). To address this question considering highly complex historical and demographic models and numerous population samples, we used new statistical inference methods called Approximate Bayesian Computation (ABC; Beaumont et al. 2002, Tavaré et al. 1997). Briefly, these methods proceed by simulating numerous genetic data sets under a given complex historic and demographic model. We then summarize the genetic information by calculating population genetics statistics for each simulated data set, and compare these summary statistics to the same statistics calculated on the real observed data. We infer *a posteriori* the distribution of the model parameters, such as divergence times and population sizes, that were used to produce the simulations (e.g. 0.1%) with summary statistics closest to our real data set.

Using ABC methods, we demonstrated for the first time that the genome-wide neutral genetic diversity of numerous Pygmy populations from Western Central Africa was much more consistent with scenarios of a common Pygmy origin than with scenarios considering an independent origin of each Pygmy population (relative posterior probability of the common origin scenario was 96 %; Verdu et al. 2009). We estimated in this study that the ancestral Pygmy population diverged from the ancestral non-Pygmy population in a remote past, roughly 50,000 - 90,000 years ago. Furthermore, we found that the ancestral Western Central African Pygmy population split much more recently, about 3,000 years ago, to give birth to the various Pygmy populations found today in Cameroon and Gabon. Finally, our genetic data further indicated that the rapid genetic diversification among Western Pygmy population during the last

3,000 years could be explained by reproductive isolation and genetic drift in each Pygmy population separately.

Importantly, Patin and colleagues further applied similar ABC methods to formally test whether Western and Eastern Pygmy groups of populations had a common or an independent origin (Patin et al. 2009). They found a likely common origin between Eastern and Western Pygmy groups roughly 20,000 years ago, consistently with previous estimates. Furthermore, they estimated that the original divergence between ancestral Pygmy and non-Pygmy populations occurred roughly 60,000 years ago. Finally, using full mitochondrial DNA sequences, Batini and colleague (2011) further explored the common or independent origin of Eastern and Western Pygmy maternal lineages and detailed scenarios of their respective past demography. These authors expanded Patin and colleagues results, finding a likely common origin of Eastern and Western Pygmy maternal lineages about 27,000 years ago, and an ancient common origin between Pygmies and non-Pygmies about 70,000 years ago (Batini et al. 2011). They also found that Pygmies likely experienced a decrease in maternal effective population sizes since about 4,000 years ago until recently, and that, conversely, non-Pygmies likely experienced a strong increase in maternal effective population size starting shortly after the original split (about 65,000 years ago) until the present day. It is worth mentioning here that all the divergence times here reported have potentially wide credibility intervals and will need to be more accurately estimated in the future using more genetic makers and individual samples⁶. A schematic population genetics based model for the peopling history of Central African Pygmy and non-Pygmy populations is presented in Figure 3.

It is difficult, if not impossible, for population geneticists to determine the external causes for the divergence and demographic changes experienced by Central African ancestral populations, especially the ancestral Pygmy/non-Pygmy original divergence about 70,000 years ago. Nevertheless, a number of archaeological and paleoclimatological data provide interesting sets of hypotheses to build a more comprehensive evolutionary framework for the divergence history in the ancestral Pygmy lineages. Patin, Batini and colleagues found that the Western-Eastern ancestral Pygmy divergence event coincided with the last Glacial Maximum. This period is characterized by a fragmentation of the forest and the formation of small forest refuges

⁶ Detailed discussions on these divergence time estimates using ABC can be found in Verdu et al. (2009), Patin et al. (2009) and Batini et al. (2011)

throughout the Congo Basin. Therefore, these authors hypothesized that the reduction of forest-covered areas potentially triggered the isolation of ancestral Western and Eastern Pygmy populations (Batini et al. 2011, Patin et al. 2009). However, paleoclimatic and paleoecological data are scarce to precisely map the fluctuations of the forest coverage in the Congo Basin over the last 30,000 years; and this hypothesis makes a number of yet unproven assumptions. In particular, it assumes that current Eastern and Western Pygmy populations settle the same areas as their ancestors and that these areas indeed formed forest refuges more than 20,000 years ago. It also assumes that Pygmy populations are and have always been confined to forest environments, which remains to be demonstrated.

Concerning the Western Congo Basin, archaeological and archaeolinguistic data respectively showed that agriculture and Bantu languages emerged about 4,000 – 5,000 years ago and rapidly spread in Central and Southern Africa (Phillipson 2005, Vansina 1995). In addition, population geneticists showed that the diffusion of Bantu languages may have been accompanied by human population migrations, as attested by the demographic expansion of non-Pygmy Bantu speaking populations and the high levels of non-Pygmy migrations found using genetic data⁷ (Berniell-Lee et al. 2009, de Filippo et al. 2011, Montano et al. 2011, Quintana-Murci et al. 2008). Pygmy and non-Pygmy populations currently share complex socioeconomic relationships that have been shown, at least in some Aka and Baka groups, to influence substantially the mobility and potential isolation of Pygmy populations (Bahuchet 1992, Hewlett et al. 1982, MacDonald and Hewlett 1999). Therefore, we proposed that the emergence of agriculture and migrations of Bantu-speaking non-Pygmies may have triggered profound social and demographic changes in the ancestral Western Pygmy population. This may in turn have led to the fragmentation and isolation of the various Pygmy populations currently settling CAR, Cameroon and Gabon over the past 3,000 years (Verdu et al. 2009). However, we did not know at the time that a recent climatic crisis occurring roughly 4,000 - 5,000 years ago resulted in the contraction of forest areas separated by vast zones of more arid savanna in Western Central Africa (Hessler et al. 2010, Lebamba et al. 2009, Maley et al. 2012). Therefore, following Patin, Batini and colleagues, and with the caution invoked above, we can further our hypothesis by

⁷ These mechanisms consistently explain the low levels of genetic differentiation found among non-Pygmy Bantu speaking populations, since such demographic expansions and migrations would homogenize the gene-pools among populations.

proposing that the agricultural and cultural revolution and/or the climatic crisis and the formation of forest refuges, may have triggered the divergence and isolation of Western Central African Pygmy populations 3,000 years ago.

II. Migration patterns among Central African populations

a. Marriage practices and gene flow among Pygmies and non-Pygmies

In both the scientific community and the non-specialized Western audience, African Pygmies are traditionally viewed as isolated from other populations, living remotely in the heart of the rainforest while agricultural non-Pygmy populations live outside forest areas. This somewhat romantic view has been frequently contradicted by the extensive work of ethnographers and anthropologists (see other chapters in this volume and Bahuchet 1992, Hewlett 1996, Joiris 2003): *i)* most Pygmy populations also often practice agriculture and eat agricultural produces on daily bases; *ii)* non-Pygmy populations practice a slash and burn agriculture inside the forest and also often eat forest goods such as game or wild honey; *iii)* numerous Pygmy groups settle in the vicinity or even inside non-Pygmy villages, if not always permanently, at least for long periods of time; and *iv)* Pygmies and non-Pygmy neighbors interact socially (including with intermarriages) and economically on daily bases. In addition, the work of Cavalli-Sforza and collaborators already identified genetic signatures of migrations between Aka Pygmies and non-Pygmies (Cavalli-Sforza 1986), further casting doubt on the representation that Pygmies are isolated from any other Central African population.

Since the late 90's population geneticists studying sex-specific genetic markers from the male inherited Y-chromosome and the female inherited mitochondrial DNA, further refined sex specific patterns of migration in Sub-Saharan Africa. In particular, Salas and colleagues found some levels of sharing of mitochondrial DNA diversity between Pygmy and non-Pygmy populations. Population geneticists attributed these patterns to traces of maternal gene flow between Pygmies and non-Pygmies through patrilocal marriage customs⁸, which would have occurred during the migrations of Bantu speaking populations (Salas et al. 2002, Seielstad et al. 1998). This hypothesis was expanded by later findings from Destro-Bisol, Batini, Quintana-Murci and their respective colleagues considering numerous Pygmy population samples (Batini

⁸ When marriage customs dictate that the bride moves to live with the husband's family after marriage.

et al. 2007, Destro-Bisol et al. 2004a, Quintana-Murci et al. 2008). Furthermore, Hammer and colleagues found that the diversity of the male inherited Y-chromosome, showing small among-population genetic differentiation, was also consistent with extensive male migrations among Sub-Saharan populations, a surprising result at odds with patrilocal customs in Central Africa (Hammer et al. 2001).

In a founding article, Destro-Bisol and colleagues (2004b) used the interdisciplinary framework of Cavalli-Sforza and Hewlett to study complex interactions between cultural and biological phenomena. They reconciled the apparent discrepancies previously found between male and female signatures of inter-population migrations by further showing how complex sociocultural behaviors could trigger movements of spouses and admixed offspring. Indeed, ethnologists previously showed that while marriages between a Pygmy male and a non-Pygmy female are often strongly prohibited, marriages between a Pygmy female and a non-Pygmy male can occur more frequently (Bahuchet and Guillaume 1982, Hewlett 1996, Joiris 2003). Nevertheless, social discriminations against the newly wedded Pygmy female that moved to the village of her in laws often lead to divorce. The Pygmy wife then returns to her community of origin, potentially with her admixed offspring. Importantly, such mobility of Pygmy females and admixed offspring in intermarriages also occurs with the death of the non-Pygmy husband. This culture-driven mobility of spouses and admixed offspring could thus explain male mediated gene-flow from non-Pygmies into Pygmy populations in a context of patrilocality and social discrimination against Pygmies.

We further used our extensive population sample set and genome-wide genetic markers to more precisely estimate admixture levels and assess the potential variability of admixture patterns across numerous Pygmy populations (Verdu et al. 2009). Results showed that all Western Central African Pygmy populations considered in our study exhibited substantial levels of asymmetric admixture from the non-Pygmy populations, hence expanding previous findings described above. Surprisingly, we found that average levels of admixture were largely variable across Pygmy populations. For instance Baka and Bezan Pygmies were significantly less admixed than Bongo populations. Using ethnographic data we observed that the heterogeneous levels of admixture found across Western Pygmy populations were inversely proportional to the strength of barriers against intermarriages: strong sociocultural prohibition of intermarriages and discriminatory endogenous representations against Pygmies could be the cause of lower genetic

admixture rates (Verdu et al. 2009). Considering Eastern Pygmy population samples, Patin and colleagues (2009), followed by Tishkoff and colleagues (2009), further found that Eastern Pygmy populations were also substantially admixed with non-Pygmies. Altogether, these findings showed that a Pygmy population with virtually no trace of admixture with non-Pygmies remains yet to be found, if such a population even exists today.

b. Dispersal behavior and reproductive isolation between neighboring Pygmy populations

We have seen that the various Pygmy populations were genetically differentiated despite sharing a recent common genetic origin in the West and a more ancient one between Eastern and Western groups of populations. The reproductive isolation is not surprising between geographically distant Eastern and Western Pygmy populations. In the light of the known exploratory mobility of Pygmy populations, such isolation is more unexpected locally, for instance between Baka and Koya neighboring Pygmy populations (Figure 1). Indeed, Pygmies are traditionally described as being extremely mobile in the forest environment (Bahuchet 1993, Hewlett 1996). Ethnographers demonstrated that this mobility was mainly due to social interactions such as search for spouses, social and ritual events among the Pygmy communities, or socio-economic interactions with non-Pygmy neighbors (Bahuchet 1992, Hewlett et al. 1982, MacDonald and Hewlett 1999). If such extended exploratory mobility translates into extended reproductive mobility, it would be surprising to identify genetic isolation signals between neighboring Pygmy populations.

When trying to assess the extent to which exploratory mobility relates to reproductive mobility or effective dispersal, namely how far children are born from the birth place of their parents, detailed demographic data on birth place of biologically-related family members are needed but are not readily available in Pygmy populations. The correlation between genetic differentiation levels and geographic distances between birth places can indirectly provide accurate estimates of effective dispersal ranges among generations. This was previously confirmed by population genetics studies of New-Guinean populations where extensive trans-generational demographic data were available (Long et al. 1987, Rousset 1997, Wood 1987). Applying these population genetics methods to Cameroonian Baka Pygmy populations, we found a significant pattern of genetic isolation by distance corresponding to very short effective dispersal ranges between 12.4 and 63.2 km² on average (Verdu et al. 2010). In other words, two

Baka individuals born far away from each other are likely to be genetically more differentiated than two individuals born close-by. Following Cavalli-Sforza and Hewlett's (1982) assumptions about the dispersal shape among Aka Pygmies, these effective dispersal ranges indicated that, on average, half of the Baka individuals were born at most between 1.7 and 3.9 km away from their parents' respective birth places (Verdu et al. 2010). Such a limited reproductive dispersal behavior, if found in other Pygmy populations, could very well explain the rapid and extensive levels of genetic differentiation found across neighboring Pygmy populations from Western Central Africa. In any case, this study showed that detailed quantitative demographic and ethnographic data on the trans-generational mobility behaviors of Pygmy populations would provide key elements to understand how and why geographically neighboring Pygmy populations are currently genetically isolated. Future population genetics work needs to be conducted to assess more precisely when such limited effective dispersal behavior started among the Baka.

III. The stature of African Pygmies

The widely different life-styles and ecology of Pygmies and non-Pygmies could have triggered different biological and morphological adaptation processes between groups (see other chapter in this volume). Understanding the differences in adult height between Pygmies and non-Pygmies best exemplifies this research paradigm (Perry and Dominy 2009).

Pygmies are found in the lower part of the adult average height distribution of African or worldwide populations. Nevertheless, the extensive work of Froment and Hiernaux (Froment 1993, Hiernaux 1974) established that there was no discontinuity in height between Central African Pygmies and non-Pygmies, some non-Pygmy populations neighboring Pygmy groups being also of short stature⁹. Therefore these authors demonstrated that considering an arbitrary threshold of height could be extremely misleading to categorize populations as Pygmies or non-Pygmies in the multi-ethnic context of Central Africa, thus contradicting Cavalli-Sforza's proposal to set the threshold for Pygmy height to 160 cm (1986). Furthermore, these authors found tremendous variance in adult height within and across Pygmy populations, ranging from

⁹ e.g. the Ba.Konjo neighboring Efe Pygmies, at the Western Ugandan border with DRC's Nord Kivu, measure less than 160 cm high on average

an average 143 cm high among Efe men in Eastern DRC, to an average 161 cm for Twa men from the Lake Tumba in Western DRC.

Based on these observations, Hiernaux proposed that the short stature of African Pygmies was due to convergent adaptation processes to ecological, parasitological and nutritional constraints in the Congo Basin forest. Cavalli-Sforza finding clues for a common origin of Pygmy populations conversely proposed that the short stature of Pygmies was inherited from a common ancestral population that had gone through an adaptation to the hot and humid environment of the equatorial forest (Cavalli-Sforza et al. 1969, Merimee et al. 1972). Indeed, thermoregulation is hard in a hot and humid environment and being short lowers the energy requirements for thermoregulation, thus providing an adaptive advantage over taller individuals. Diamond further proposed that Pygmies short stature could reflect an adaptation to hunting and gathering in the dense rainforest environment (Diamond 1991). In this hypothesis, being shorter would allow easier mobility in the rainforest and thus lower energy expenditure while looking for food. Furthermore, Bailey proposed that Pygmies short stature allowed reduced caloric requirements, which could be advantageous in the nutritionally scarce forest environment (Bailey 1991, Bailey et al. 1989). More recently, Migliano and colleagues (2007) proposed that, in the context of high mortality in the equatorial forest, it could be advantageous for the survival of the population to start reproduction earlier, which would in turn result in a reduction in stature of the population. However, none of these hypotheses was formally proven to be true, and all of them were challenged by other anthropology and ethnography work. Furthermore, anthropologists are still debating about whether the short stature of African Pygmies is fully due to environmental phenotypic plasticity rather than to stem from a biological adaptation to natural selection pressures in a challenging environment (Bahuchet 1988, Bahuchet et al. 1991, Froment 1993, Becker et al. 2010, Perry and Dominy 2009).

Physiologists attributed the short stature of Pygmies as compared to non-Pygmies to potential differences between populations in the metabolic pathway determining in part cell and tissue growth: the Growth Hormone Receptor (GHR) - Insulin Growth Factor (IGF) pathway (Merimee et al. 1972, Merimee et al. 1987, Rimoin et al. 1969). Recently, Bozzola and colleagues (2009) found that African Pygmies had reduced expression levels of certain hormones and receptors of the GHR – IGF pathway as compared to non-Pygmies. However, several studies focusing on the genes encoding some of the proteins of this pathway failed to identify genetic

mutations significantly differentiating Pygmies from non-Pygmies (Bowcock and Sartorelli 1990).

Most these studies implicitly or explicitly assumed the common origin of Pygmy populations, and hence a possible common inheritance of the genetic determinant of height from the ancestral Pygmy population. Although we recently proved that genetic data advocated indeed for a common origin of Central African Pygmy populations, further issues concerning the evolution of height emerge. In the lack of ancient human remains in Central Africa, we do not know if an ancient population of particularly short stature existed in the region or not. Therefore, we do not know if Pygmies became shorter in the course of evolution, or if neighboring non-Pygmies conversely became taller. This major issue has been somewhat overlooked by some biological anthropologists. However, the answer could fundamentally change the way the question of the genetic determination of Central African differences in stature and the identification of potential signatures of natural selection influencing this phenotype should be addressed.

In this context, Becker and colleagues (2011) recently looked for indirect evidences of genetic determination of the differences in height between Pygmies and non-Pygmies. Using numerous Pygmy and neighboring non-Pygmy population samples, categorized as such based on cultural criteria disregarding *a priori* height information, these authors found a significant positive correlation between individual's height and levels of genetic admixture among Pygmies and non-Pygmies. A Central African individual categorized as Pygmy based on cultural criteria and found to be highly genetically admixed with non-Pygmies was likely to be significantly taller than a less admixed Pygmy individual. This showed for the first time that yet unidentified genetic factors could be involved in determining height differences between Pygmies and non-Pygmies. This study highlighted that the diversity of Pygmy populations should imperatively be considered and genetic substructures, such as admixture patterns, controlled for when trying to identify genetic determinants associated with height among Central Africans. Recently, Jarvis and collaborators, following the path opened by Becker and collaborators, released a genome-wide analysis using more than a million genetic markers to identify mutations determining height differences among Central Africans (Jarvis et al. 2012). These authors identified a number of mutations spread out across the genome showing signatures of differential adaptation between Pygmies and non-Pygmies. In particular, they found that genomic regions coding for proteins

involved in immunological functions were enriched in mutations strongly differentiating Pygmies and non-Pygmies and showing signatures of differential natural selection as compared to the rest of the genome. These results were very recently further confirmed by the study of whole genome sequences of 15 individuals from three Cameroonian Pygmy populations (Lachance et al. 2012). Therefore, Jarvis, Lachance and collaborators proposed that the short stature of Central African Pygmies may be resulting from an adaptation to challenging parasitological environments. However, they could not identify genetic mutations significantly associated to height differences in Central Africa, mainly due to a lack of statistical power when considering a very limited number of individuals from both groups of populations. These studies nevertheless paved the way for future promising studies trying to solve the long-standing and still unsolved mystery of Central African Pygmies and non-Pygmies differential stature.

Perspectives and conclusions

Perspectives on the origins of Central African populations

Population genetics approaches unveiled the common origin of numerous culturally, morphologically and genetically diverse Pygmy populations and further proposed a time-line for the various divergence events among Central African populations. However, numerous questions remain unsolved. First, since population geneticists focused on the history of genes carried by individuals, the historical peopling model proposed is not geographically rooted. Indeed, we still do not know where the ancestral Pygmy and non-Pygmy populations settled, or when Pygmy and non-Pygmy populations first settled in the locations that they currently occupy. Moreover, while numerous samples from various regions of Sub-Saharan Africa are currently available, Central Africa is still overall poorly represented in population genetics databases, in particular Southern Sudan, CAR, North-Western Uganda, Rwanda, Burundi, and, most importantly, DRC. Second, the detailed divergence history of the various Pygmy populations remains unknown: the population genetics models here presented could not assess, for instance, whether the Cameroonian Baka Pygmies diverged from the common ancestral Western Pygmy population before or after the Bedzan. Furthermore, the divergence history of existing Eastern Pygmy populations has never been precisely assessed. Finally, the studies described here highlight a fundamental characteristic of the genetics of African Pygmies: the substantial levels of genetic differentiation among Pygmy populations. In this context, it is difficult to predict how additional

samples from other Pygmy populations such as the Twa from Lake Tumba in Western DRC, the Asua from North-Eastern DRC or the Cwa from Southern DRC, will modify our current understanding of the peopling of Central Africa.

Perspectives on culture/gene interactions among Pygmies and non-Pygmies

Determining since when Pygmies and non-Pygmy neighbors share complex socioeconomic relationship has been an important debate animating the ethnologist community in the past decades. The main underlying question is whether the social interactions pre-dated the beginning of the Western colonization of Central Africa in the 17th century or whether the profound social changes triggered by these colonization events subsequently provoked the emergence of new social interactions between Pygmies and non-Pygmies. Although not all sociocultural interactions lead to intermarriages resulting in genetic admixture between existing populations, estimating the time of the admixture events could at least provide a conservative lower bound for the historical onset of social interactions between communities. Finally, the complex social behaviors triggering the mobility of spouses and admixed offspring proposed by population geneticists for explaining observed admixture patterns remain to be precisely quantified in the field. Indeed, patrilocality also implies that children live with the husband family. If such behavior was strongly prevalent in Pygmy to non-Pygmy marriages and separations, an alternative explanation of the genetic admixture patterns here found should be considered. Therefore, the mobility of admixed Pygmy/non-Pygmy children in intermarriages, as well as their social status and reproductive success in either community respectively, imperatively needs to be further explored.

Concluding remarks

In the last 50 years, population genetics studies rooted in the interdisciplinary work conducted by Cavalli-Sforza, Hewlett and collaborators tremendously increased our understanding of the peopling history and biological evolution of Central African Pygmy and non-Pygmy populations. In particular, they highlighted extremely complex patterns of genetic diversity and admixture among Pygmies and non-Pygmies potentially determined by marriage behaviors. This demonstrates the importance of considering the diversity of populations designated under the umbrella term Pygmy in order to understand the evolution of Central

African populations. Following in the footsteps of ethnologists and anthropologists, population geneticists further deconstructed the pregnant myths and wide-spread misconceptions surrounding Central African Pygmy populations. Population geneticists showed that *i)* despite sharing a common origin, Pygmy populations are not biologically homogeneous but are substantially genetically differentiated from one another; *ii)* Pygmy populations are not isolated from other African populations but are all substantially admixed genetically with non-Pygmy neighbors; *iii)* Despite an extensive exploratory mobility, at least one Pygmy group has very reduced effective dispersal ranges, a demographic behavior that could trigger reproductive isolation among neighboring Pygmy populations. Finally, population geneticists showed that the differential stature between Pygmies and non-Pygmies was at least partly due to complex multifactorial genetic factors as opposed to be caused exclusively by environmental factors experienced by Pygmies during growth. These genetic factors nevertheless yet remain to be precisely identified.

While the different approaches and definitions of what is “a population” can create tremendous divisions between geneticists, anthropologists and ethnologists, the example of the complex Pygmy/non-Pygmy categorization of Central African populations shows that successful interdisciplinary efforts establishing common grounds for studying the same groups of individuals from largely different points of view can be infinitely fruitful. Indeed, interdisciplinary approaches rooted in joint field work allowed not only to rediscover and revisit unsolved questions, but also allowed to build a historical and evolutionary framework for studying with a novel eye the cultural and biological evolution of Central African populations.

TABLE

Table 1. Pygmy and non-Pygmy population samples in some recent population genetics studies. Pygmies and non-Pygmies are categorized as such based on numerous cultural criteria following anthropology and ethnology studies (see Introduction). Recent population genetics studies studying autosomal and mitochondrial DNA (mtDNA) genetic data are given separately in two different columns. Linguistic affiliations were determined following the classification in Greenberg 1966 and Guthrie 1967. “*nd*” indicates when no data was available. The column “Sample sizes used for this study” indicates the number of family unrelated individuals considered for the original population genetics analyses of genetic differentiation performed in this chapter (see section *I.a.* and Figure 2). We studied the distribution of genetic diversity within and among Central African populations using 26 independent neutral autosomal microsatellite markers that is 26 locations in the genome (not sex-specific) far from one another and far from any known or predicted genes. These genomic regions exhibit known mutations genetically differentiating individuals and potentially populations. Original publications corresponding to the genetic data here used are given between parentheses. The mention “*This study*” indicates that genetic data concerning these populations are reported in this chapter for the first time and were obtained following the protocol and normalization procedures described in Verdu et al. (2009) and Becker et al. (2010). Genotype data and technical aspects of the genotyping and normalization procedures are available upon demand to the author. French, USA, Ugandan, CAR, Gabonese and Cameroonian official ethical and research review boards delivered appropriate research and sampling authorizations for this study. DNA was originally extracted from blood or saliva samples obtained from informed voluntary donors according to standard protocols. Oral, video-recorded and/or written informed consents were provided by each donor.

Table 1.

	Population Name	Code	Country	Linguistic Family	Sampling	Autosomal Data References	mtDNA References	Sample sizes used for this study
Pygmies	Bezan	BEZ	Cameroon	Bantoïd Non-Bantu, Tikar	A. Froment, P. Verdu	Verdu et al. 2009; Tishkoff et al. 2009	Quintana-Murci et al. 2008	29 (Verdu et al. 2009)
	Kola	KOL	Cameroon	Bantu, A80	A. Froment	Verdu et al. 2009; Patin et al. 2009; Tishkoff et al. 2009	Quintana-Murci et al. 2008	31 (Verdu et al. 2009)
	Central Baka	CBK	Cameroon	Adamawa-Ubangian, Ngbaka	A. Froment	Verdu et al. 2009; Patin et al. 2009; Tishkoff et al. 2009	Quintana-Murci et al. 2008	29 (Verdu et al. 2009)
	Eastern Baka	EBK	Cameroon	Adamawa-Ubangian, Ngbaka	A. Froment	Verdu et al. 2009	<i>nd</i>	29 (Verdu et al. 2009)
	Southern Baka	SBK	Cameroon	Adamawa-Ubangian, Ngbaka	A. Froment	Verdu et al. 2009	Quintana-Murci et al. 2008	29 (Verdu et al. 2009)
	Gabonese Baka	GBK	Gabon	Adamawa-Ubangian, Ngbaka	J-M Hombert, L. Van Der Veen	Verdu et al. 2009; Patin et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Koya	KOY	Gabon	Bantu, B20	S. Le Bomin	Verdu et al. 2009	Quintana-Murci et al. 2008	29 (Verdu et al. 2009)
	Eastern Bongo	EBG	Gabon	Bantu, B70	P. Verdu	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Southern Bongo	SBG	Gabon	Bantu, B30-B50	P. Verdu	Verdu et al. 2009	<i>nd</i>	30 (Verdu et al. 2009)
	Babinga	BIN	PRC	<i>nd</i>	<i>nd</i>	<i>nd</i>	Batini et al. 2007; 2011	<i>nd</i>
	Aka	AKA	CAR	Bantu, C10	B. Hewlett	<i>This study</i>	<i>nd</i>	27 (<i>This study</i>)
	Biaka	BIA	CAR	Bantu C10	L.L. Cavalli-Sforza, B. Hewlett	Rosenberg et al. 2002; Patin et al. 2009; Tishkoff et al. 2009	Destro-Bisol et al. 2004; Quintana-Murci et al. 2008; Batini et al. 2011	36 (Rosenberg et al. 2002)
	Mbenzele Aka	MBE	CAR	Bantu C10 ?	<i>nd</i>	Destro-Bisol et al. 2004	Destro-Bisol et al. 2004; Batini et al. 2007; 2011	<i>nd</i>
	Nsua	NSU	Uganda	Sudanic ?	P. Verdu	<i>This study</i>	<i>nd</i>	18 (<i>This study</i>)
	Mbuti	MBU	DRC	Sudanic, Mangbutu-Efe/Bantu ?	L.L. Cavalli-Sforza, B. Hewlett	Rosenberg et al. 2002; Patin et al. 2009; Tishkoff et al. 2009	Destro-Bisol et al. 2004; Quintana-Murci et al. 2008; Batini et al. 2011	15 (Rosenberg et al. 2002)

	Northern Twa	NTW	Rwanda	<i>nd</i>	<i>nd</i>	Patin et al. 2009	<i>nd</i>	<i>nd</i>
	Southern Twa	STW	Rwanda	<i>nd</i>	<i>nd</i>	Patin et al. 2009	<i>nd</i>	<i>nd</i>
Non-Pygmies	Tikar	TIK	Cameroon	Bantoïde Non-Bantu, Tikar	A. Froment	Verdu et al. 2009; Tishkoff et al. 2009	<i>nd</i>	30 (Verdu et al. 2009)
	Nzime	NZI	Cameroon	Bantu, A80	A. Froment	Verdu et al. 2009; Tishkoff et al. 2009	<i>nd</i>	31 (Verdu et al. 2009)
	Bangando	BGD	Cameroon	Adamawa-Ubangian, Bangandu, Gbaya	A. Froment	Verdu et al. 2009	<i>nd</i>	30 (Verdu et al. 2009)
	Ngumba	NGB	Cameroon	Bantu, A80	A. Froment	Verdu et al. 2009; Patin et al. 2009; Tishkoff et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Ewondo	EWD	Cameroon	Bantu, A70	A. Froment	Verdu et al. 2009	Quintana-Murci et al. 2008	24 (Verdu et al. 2009)
	Fang	CFG	Cameroon	Bantu, A70	A. Froment	Verdu et al. 2009; Tishkoff et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Fang	GFG	Gabon	Bantu, A70	J-M Hombert, L. Van Der Veen	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Akele (Bongomo)	AKL	Gabon	Bantu, B20	S. Le Bomin	Verdu et al. 2009	<i>nd</i>	13 (Verdu et al. 2009)
	Teke	TEK	Gabon	Bantu, B70	J-M Hombert, L. Van Der Veen	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Nzebi	NZE	Gabon	Bantu, B50	J-M Hombert, L. Van Der Veen	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Kota	KOT	Gabon	Bantu, B20	J-M Hombert, L. Van Der Veen	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Tsogho	TSG	Gabon	Bantu, B30	J-M Hombert, L. Van Der Veen	Verdu et al. 2009	Quintana-Murci et al. 2008	30 (Verdu et al. 2009)
	Konjo	KJO	Uganda	Bantu, ?	P. Verdu	<i>This study</i>	<i>nd</i>	28 (<i>This study</i>)

FIGURE LEGENDS

Figure 1. Location of Pygmy and non-Pygmy population samples in recent population genetics studies focusing on the peopling history of Central Africa.

See Table 1 for population codes and publication references. Pygmy and non-Pygmy categories were established using numerous cultural criteria following anthropology and ethnology studies (see Introduction). Peopling areas on the map were inferred from ethnographic field work and presented in Bahuchet 2012.

Figure 2. Two-dimensional multidimensional scaling representation of Central African population pairwise genetic differentiation levels.

Pairwise levels of genetic differentiation between populations were calculated as F_{ST} (Weir and Cockerham 1984) using 26 independent tetranucleotide microsatellite markers (see Table 1). Non-significant F_{ST} values based on 10,000 permutations of individuals among populations were set to 0. Pairwise F_{ST} values are represented in two dimensions using a metric multidimensional scaling approach. To evaluate how well the two-dimensional plot represented the pairwise genetic differentiation levels, we calculated the Spearman correlation between Euclidean distances among pairs of populations on the plot and the corresponding pairwise F_{ST} levels of genetic differentiation. We obtained a value of Spearman ρ equal to 0.84 indicating a very high correlation between pairwise distances on the plot and the corresponding genetic differentiation levels. Pygmy populations are represented in red and non-Pygmies in blue. See Table 1 for population codes, sample sizes and publication references.

Figure 3. Synthetic model for the genetic origins of Central Pygmy and non-Pygmy populations. This historical peopling model of Central Africa is based mainly on population genetics studies and Approximate Bayesian Computation approaches to reconstruct the evolutionary history of Central African genetic diversity by Verdu et al. (2009), Patin et al. (2009) and Batini et al. (2011).

Figure 1.

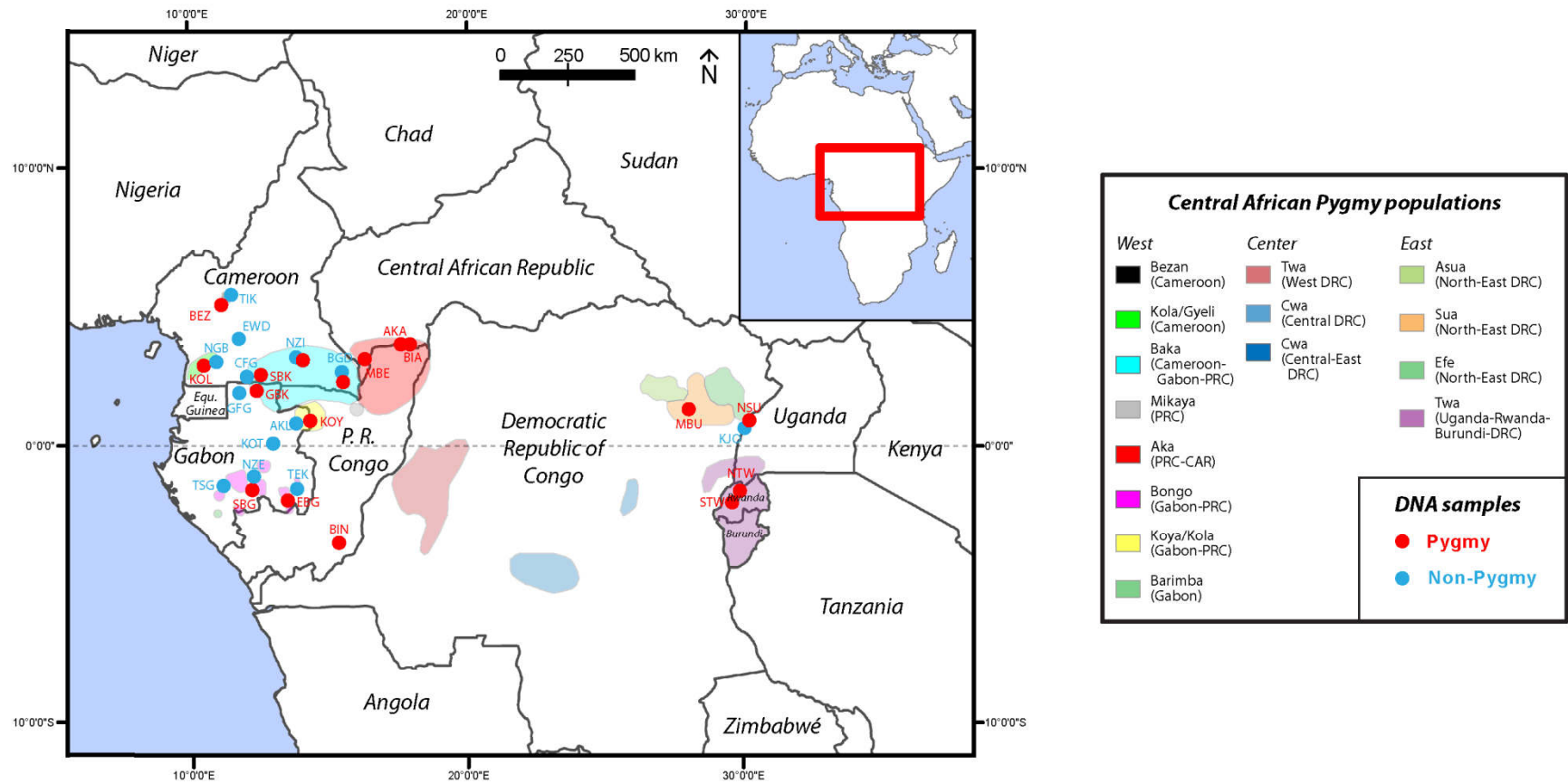


Figure 2.

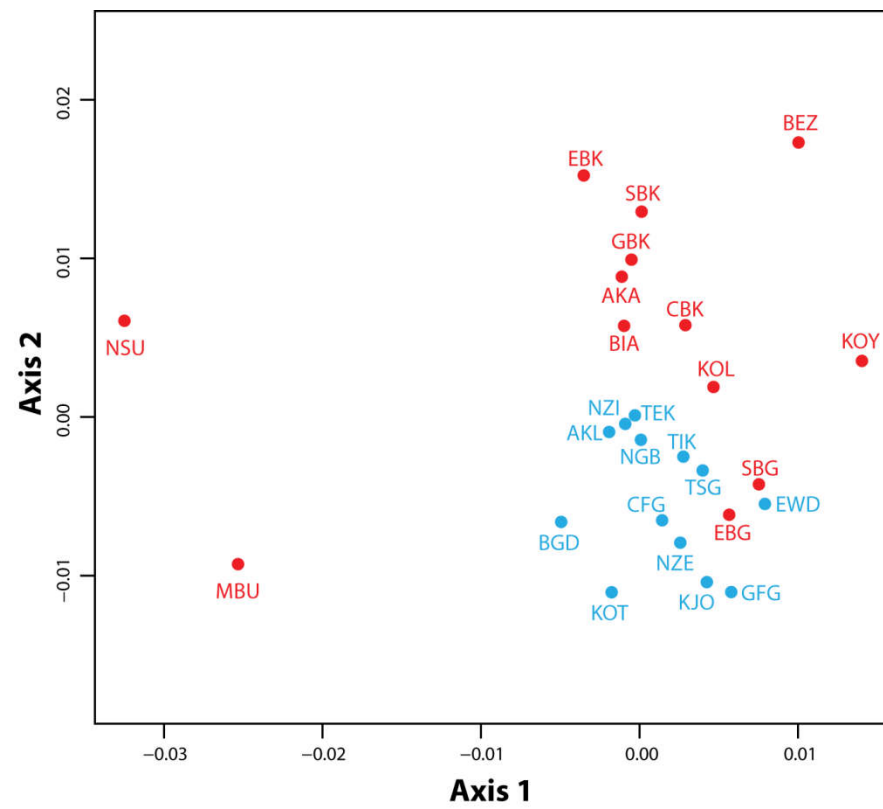
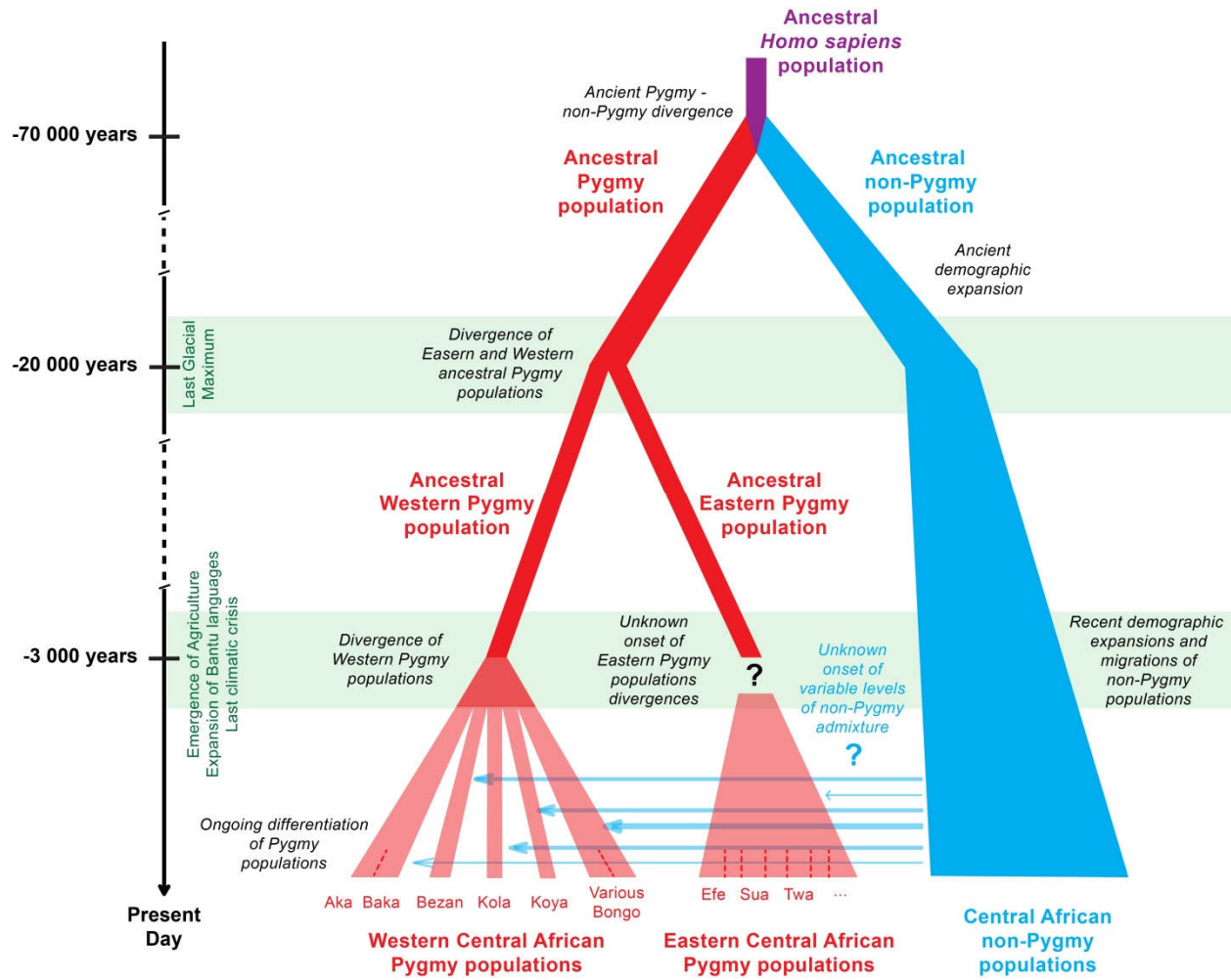


Figure 3.



REFERENCES

- Bahuchet, S. 1988. "Food supply uncertainty among the Aka Pygmies (Lobaye, C.A.R.)," in *Coping with Uncertainty in food supply*. Edited by I. d. Garine and G. A. Harrison, pp. 118-149. Oxford: Oxford University Press.
- . 1992. "Spatial mobility and access to the resources among the African Pygmies," in *Mobility and territoriality. Social and spatial boundaries among foragers, fishers, pastoralists and peripatetics*. Edited by Casimir and Rao, pp. 205-257. N.Y./Oxford: Berg.
- . 1993. L'invention des Pygmées. *Cahiers d'Etudes africaines* 33:153-181.
- . 2012. Changing language, remaining pygmy. *Hum Biol* 84:11-43.
- Bahuchet, S., and H. Guillaume. 1982. *Aka-farmer relations in the Northwest Congo Basin*. In: *Politics and History in Band Societies*. Cambridge/Paris: Cambridge University Press/M S H.
- Bahuchet, S., D. McKey, and I. de Garine. 1991. Wild yams revisited: is independence from agriculture possible for rain forest hunter-gatherers? *Human Ecology* 19:213-243.
- Bailey, R. C. 1991. The comparative growth of Efe pygmies and African farmers from birth to age 5 years. *Ann Hum Biol* 18:113-20.
- Bailey, R. C., G. Head, M. Tenike, B. Owen, R. Rechtman, et al. 1989. Hunting and gathering in tropical rainforest: is it possible? *American Anthropologist* 91:59-82.
- Batini, C., V. Coia, C. Battaglia, J. Rocha, M. M. Pilkington, et al. 2007. Phylogeography of the human mitochondrial L1c haplogroup: genetic signatures of the prehistory of Central Africa. *Mol Phylogenet Evol* 43:635-44.
- Batini, C., J. Lopes, D. M. Behar, F. Calafell, L. B. Jorde, et al. 2011. Insights into the demographic history of African Pygmies from complete mitochondrial genomes. *Mol Biol Evol* 28:1099-110.
- Beaumont, M. A., W. Zhang, and D. J. Balding. 2002. Approximate Bayesian computation in population genetics. *Genetics*. 162:2025-35.
- Becker, N. S., P. Verdu, A. Froment, S. Le Bomin, H. Pagezy, et al. 2011. Indirect evidence for the genetic determination of short stature in African Pygmies. *Am J Phys Anthropol* 145:390-401.
- Becker, N. S. A., P. Verdu, B. Hewlett, and S. Pavard. 2010. Can life history trade-offs explain the evolution of short stature in human pygmies? A response to Migliano and colleagues. *Human Biology* vol. 82.
- Berniell-Lee, G., F. Calafell, E. Bosch, E. Heyer, L. Sica, et al. 2009. Genetic and demographic implications of the Bantu expansion: insights from human paternal lineages. *Mol Biol Evol* 26:1581-9.
- Bowcock, A., and V. Sartorelli. 1990. Polymorphism and mapping of the IGF1 gene, and absence of association with stature among African Pygmies. *Hum Genet* 85:349-54.
- Bozzola, M., P. Travaglino, N. Marziliano, C. Meazza, S. Pagani, et al. 2009. The shortness of Pygmies is associated with severe under-expression of the growth hormone receptor. *Mol Genet Metab* 98:310-3.
- Campbell, M. C., and S. A. Tishkoff. 2010. The evolution of human genetic and phenotypic variation in Africa. *Curr Biol* 20:R166-73.
- Cann, H. M., C. de Toma, L. Cazes, M. F. Legrand, V. Morel, et al. 2002. A human genome diversity cell line panel. *Science* 296:261-2.
- Cavalli-Sforza, L. I. 1986. *African Pygmies*. Orlando, FL: Academic Press.
- Cavalli-Sforza, L. L. 2005. The Human Genome Diversity Project: past, present and future. *Nat Rev Genet* 6:333-40.
- Cavalli-Sforza, L. L., S. P. Daiger, and D. P. Rummel. 1977. Detection of genetic variation with radioactive ligands. I. Electrophoretic screening of plasma proteins with a selected panel of compounds. *Am J Hum Genet* 29:581-92.

- Cavalli-Sforza, L. L., and M. W. Feldman. 2003. The application of molecular genetic approaches to the study of human evolution. *Nat Genet.* 33 Suppl:266-75.
- Cavalli-Sforza, L. L., and B. Hewlett. 1982. Exploration and mating range in African Pygmies. *Ann. Human Genetics* 46:257-270.
- Cavalli-Sforza, L. L., L. A. Zonta, F. Nuzzo, L. Bernini, W. W. de Jong, et al. 1969. Studies on African Pygmies. I. A pilot investigation of Babinga Pygmies in the Central African Republic (with an analysis of genetic distances). *Am J Hum Genet* 21:252-74.
- Cornelissen, E. 2002. Human responses to changing environments in Central Africa between 40,000 and 12,000 B.P. *Journal of World Prehistory* 16:197-235.
- Daiger, S. P., and L. L. Cavalli-Sforza. 1977. Detection of genetic variation with radioactive ligands. II. Genetic variants of vitamin D-labeled group-specific component (Gc) proteins. *Am J Hum Genet* 29:593-604.
- de Filippo, C., C. Barbieri, M. Whitten, S. W. Mpoloka, E. D. Gunnarsdottir, et al. 2011. Y-chromosomal variation in sub-Saharan Africa: insights into the history of Niger-Congo groups. *Mol Biol Evol* 28:1255-69.
- DeGiorgio, M., M. Jakobsson, and N. A. Rosenberg. 2009. Explaining worldwide patterns of human genetic variation using a coalescent-based serial founder model of migration outward from Africa. *Proc Natl Acad Sci U S A* 106:16057-62.
- Destro-Bisol, G., I. Boschi, A. Caglia, S. Tofanelli, V. Pascali, et al. 2000. Microsatellite variation in Central Africa: an analysis of intrapopulational and interpopulational genetic diversity. *Am J Phys Anthropol* 112:319-37.
- Destro-Bisol, G., V. Coia, I. Boschi, F. Verginelli, A. Caglia, et al. 2004a. The analysis of variation of mtDNA hypervariable region 1 suggests that Eastern and Western Pygmies diverged before the Bantu expansion. *American Naturalist* 163:212-226.
- Destro-Bisol, G., F. Donati, V. Coia, I. Boschi, F. Verginelli, et al. 2004b. Variation of female and male lineages in sub-Saharan populations: the importance of sociocultural factors. *Mol Biol Evol.* 21:1673-82.
- Diamond, J. M. 1991. Anthropology. Why are pygmies small? *Nature* 354:111-2.
- Du Chaillu, P. B., and R. Owen. 1867. *A journey to Ashango-Land: and further penetration into equatorial Africa*. New York,: D. Appleton and company.
- Excoffier, L., P. E. Smouse, and J. M. Quattro. 1992. Analysis of molecular variance inferred from metric distances among DNA haplotypes: application to human mitochondrial DNA restriction data. *Genetics.* 131:479-91.
- Froment, A. 1993. Adaptation biologique et variation dans l'espèce humaine : le cas des Pygmées d'Afrique. *Bulletin et Mémoires de la Société d'anthropologie de Paris* 5:417-448.
- Greenberg, J. H. 1966. *The languages of Africa*. Bloomington/The Hague: Indiana University/Mouton & Co.
- Guthrie, M. 1967. *Comparative Bantu; an introduction to the comparative linguistics and prehistory of the Bantu languages*. Farnborough,: Gregg.
- Hammer, M. F., T. M. Karafet, A. J. Redd, H. Jarjanazi, S. Santachiara-Benerecetti, et al. 2001. Hierarchical patterns of global human Y-chromosome diversity. *Mol Biol Evol.* 18:1189-203.
- Hessler, I., L. Dupont, R. Bonnefille, H. Behling, C. Gonzalez, et al. 2010. Millennial-scale changes in vegetation records from tropical Africa and South America during the last glacial. *Quaternary Science Reviews* 29:2882-2899.
- Hewlett, B. 1996. "Cultural diversity among African Pygmies," in *Cultural diversity among twentieth-century foragers. An African perspective*, vol. 1. Edited by S. Kent, pp. 215-244. Cambridge: Cambridge University Press.

- Hewlett, B., J. Van de Koppel, and L. L. Cavalli-Sforza. 1982. Exploration ranges of Aka Pygmies of the Central African Republic. *Man* 17:418-430.
- Hiernaux, J. 1974. *The people of Africa*. London: Weidenfeld and Nicolson.
- Jarvis, J. P., L. B. Scheinfeldt, S. Soi, C. Lambert, L. Omberg, et al. 2012. Patterns of ancestry, signatures of natural selection, and genetic association with stature in Western African pygmies. *PLoS Genet* 8:e1002641.
- Joiris, D. V. 2003. The framework of Central African hunter-gatherers and neighbouring societies. *African Study Monographs, Suppl.* 28:57-79.
- Lachance, J., B. Vernot, C. C. Elbers, B. Ferwerda, A. Froment, et al. 2012. Evolutionary history and adaptation from high-coverage whole-genome sequences of diverse african hunter-gatherers. *Cell* 150:457-69.
- Lebamba, J., A. Ngomanda, A. Vincens, D. Jolly, C. Favier, et al. 2009. Central African biomes and forest succession stages derived from modern pollen data and plant functional types. *Climate of the Past* 5:403-429.
- Long, J. C., P. E. Smouse, and J. W. Wood. 1987. The allelic correlation structure of Gainj- and Kalam-speaking people. II. The genetic distance between population subdivisions. *Genetics* 117:273-83.
- Macaulay, V., C. Hill, A. Achilli, C. Rengo, D. Clarke, et al. 2005. Single, rapid coastal settlement of Asia revealed by analysis of complete mitochondrial genomes. *Science*. 308:1034-6.
- MacDonald, D. H., and B. S. Hewlett. 1999. Reproductive interests and forager mobility. *Current Anthropology* 40:501-523.
- Maley, J., P. Giresse, C. Doumenge, and C. Favier. 2012. Comment on "Intensifying weathering and land use in Iron Age Central Africa". *Science* 337:1040; author reply 1040.
- Mercader, J. Editor. 2003. *Under the canopy. The archaeology of tropical rain forests*. New Brunswick: Rutgers University Press.
- Merimee, T. J., D. L. Rimoïn, and L. L. Cavalli-Sforza. 1972. Metabolic studies in the African pygmy. *J Clin Invest* 51:395-401.
- Merimee, T. J., J. Zapf, B. Hewlett, and L. L. Cavalli-Sforza. 1987. Insulin-like growth factors in pygmies. The role of puberty in determining final stature. *N Engl J Med* 316:906-11.
- Migliano, A. B., L. Vinicius, and M. M. Lahr. 2007. Life history trade-offs explain the evolution of human pygmies. *Proc Natl Acad Sci U S A* 104:20216-9.
- Montano, V., G. Ferri, V. Marcari, C. Batini, O. Anyaele, et al. 2011. The Bantu expansion revisited: a new analysis of Y chromosome variation in Central Western Africa. *Mol Ecol* 20:2693-708.
- Patin, E., G. Laval, L. B. Barreiro, A. Salas, O. Semino, et al. 2009. Inferring the demographic history of african farmers and pygmy hunter-gatherers using a multilocus resequencing data set. *PLoS Genet* 5:e1000448.
- Perry, G. H., and N. J. Dominy. 2009. Evolution of the human pygmy phenotype. *Trends Ecol Evol* 24:218-25.
- Phillipson, D. W. 2005. *African archaeology*, 3rd edition. Cambridge: Cambridge University Press.
- Quintana-Murci, L., H. Quach, C. Harmant, F. Luca, B. Massonnet, et al. 2008. Maternal traces of deep common ancestry and asymmetric gene flow between Pygmy hunter-gatherers and Bantu-speaking farmers. *Proc Natl Acad Sci U S A*. 105:1596-601.
- Quintana-Murci, L., O. Semino, H. J. Bandelt, G. Passarino, K. McElreavey, et al. 1999. Genetic evidence of an early exit of Homo sapiens sapiens from Africa through eastern Africa. *Nat Genet*. 23:437-41.
- Rimoïn, D. L., T. J. Merimee, D. Rabinowitz, L. L. Cavalli-Sforza, and V. A. McKusick. 1969. Peripheral subresponsiveness to human growth hormone in the African pygmies. *N Engl J Med* 281:1383-8.
- Rosenberg, N. A., J. K. Pritchard, J. L. Weber, H. M. Cann, K. K. Kidd, et al. 2002. Genetic structure of human populations. *Science*. 298:2381-5.

- Rousset, F. 1997. Genetic differentiation and estimation of gene flow from F-statistics under isolation by distance. *Genetics* 145:1219-1228.
- Salas, A., M. Richards, T. De la Fe, M. V. Lareu, B. Sobrino, et al. 2002. The making of the African mtDNA landscape. *Am J Hum Genet.* 71:1082-111.
- Santachiara-Benerecetti, A. S., M. Beretta, M. Negri, G. Ranzani, G. Antonini, et al. 1980. Population genetics of red cell enzymes in Pygmies: a conclusive account. *Am J Hum Genet* 32:934-54.
- Schweinfurth, G. 1873. *Im Herzen von Afrika*. Leipzig: Brockhaus.
- Seielstad, M. T., E. Minch, and L. L. Cavalli-Sforza. 1998. Genetic evidence for a higher female migration rate in humans. *Nat Genet.* 20:278-80.
- Tavaré, S., D. J. Balding, R. C. Griffiths, and P. Donnelly. 1997. Inferring coalescence times from DNA sequence data. *Genetics* 145:505-518.
- Tishkoff, S. A., F. A. Reed, F. R. Friedlaender, C. Ehret, A. Ranciaro, et al. 2009. The genetic structure and history of Africans and African Americans. *Science* 324:1035-44.
- Vansina, J. 1995. New linguistic evidence and 'the Bantu expansion'. *Journal of African History* 36:173-195.
- Verdu, P., F. Austerlitz, A. Estoup, R. Vitalis, M. Georges, et al. 2009. Origins and genetic diversity of pygmy hunter-gatherers from Western Central Africa. *Curr Biol* 19:312-8.
- Verdu, P., R. Leblois, A. Froment, S. Thery, S. Bahuchet, et al. 2010. Limited dispersal in mobile hunter-gatherer Baka Pygmies. *Biol Lett* 6:858-61.
- Weir, B. S., and C. C. Cockerham. 1984. Estimating F-Statistics for the Analysis of Population-Structure. *Evolution* 38:1358-1370.
- Wood, J. W. 1987. The Genetic Demography of the Gainj of Papua-New-Guinea .2. Determinants of Effective Population-Size. *American Naturalist* 129:165-187.