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The Population History of the Philippines: A Genetic Overview

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Professorial Address

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The Population History of the Philippines A Genetic Overview

This professorial address synthesizes the findings of several studies on the genetics of Philippine population groups and presents them in a manner accessible to nonspecialists. These studies examined different types of DNA (Y-chromosome, mitochondrial, and autosomal) samples contributed by participants in twelve regional centers and nineteen ethnolinguistic groups. The data reveal (a) genetic affinities with different groups in the Asia-Pacific region, (b) genetic signals of ancient demographic events, (c) an ancient genetic history of about 5,000–50,000 years ago, and (d) a complex population genetic structure. This genetic overview has implications for the study of human history, forensics, and medicine.

KEYWORDS: DNA • GENETICS • HISTORY • DEMOGRAPHY • POPULATION STRUCTURE

The Philippines is popularly described as a diverse country not only in terms of its geography but also in terms of its peoples, languages, and cultures. Over 100 different ethnolinguistic groups exist, each of which is considered a separate cultural entity. There is also diversity in the foreign influences on the country that emanated from different parts of Asia as well as from Europe and the Americas. The origins, history, and diversity of the Filipino people make the Philippines relevant in the study of human diversity, population history, and human origins in the Asia-Pacific region.¹

In terms of human origins, the “Waves of Migration Theory” suggests that the Philippines was peopled through a series of human migrations that reached the Philippine archipelago as early as 250,000 years ago (ya) up to the Christian era around 200–300 ya (Beyer 1950; Jocano 1998) (fig. 1). The ancient migrations traversed land bridges that were exposed due to shallow sea water levels and which connected the Sunda shelf with Palawan and the Sulu Archipelago and with Mindanao via the Celebes Sea (Beyer 1950; Scott 1984; Jocano 1998). However, current research in archaeology and linguistics point to two major demographic events in the Asia-Pacific region that could have contributed to the peopling of the Philippines (fig. 2). The initial peopling of the Asia-Pacific region is estimated to have occurred about 50,000–70,000 ya (O’Connell and Allen 2004; Barker et al. 2007). Philippine fossil records show human remains as old as 36,000–68,000 ya (Détroit et al. 2004; Mijares et al. 2010). The overlap in ages suggests human occupation in the Philippines as early as the initial peopling of the Asia-Pacific region.

The spread of Austronesian languages—the Austronesian expansion—is said to have originated in Taiwan, about 4,000–7,000 ya (Bellwood 1984; Gray et al. 2009) (fig. 2). Commonly known as the Out-of-Taiwan hypothesis, the Austronesian speakers first migrated through the Philippines before moving on to other parts of the Asia-Pacific. The influence of this demographic event is evidenced by the fact that all Philippine languages belong to the Austronesian language family (Lewis et al. 2015).

The demographic structure of the current Philippine population can be described in layers. Over 100 different ethnolinguistic groups living in their respective communities across the archipelago make up the base layer. Several ethnolinguistic communities make up towns; several towns make up over 100 cities, which in turn comprise about eighty provinces. Philippine provinces

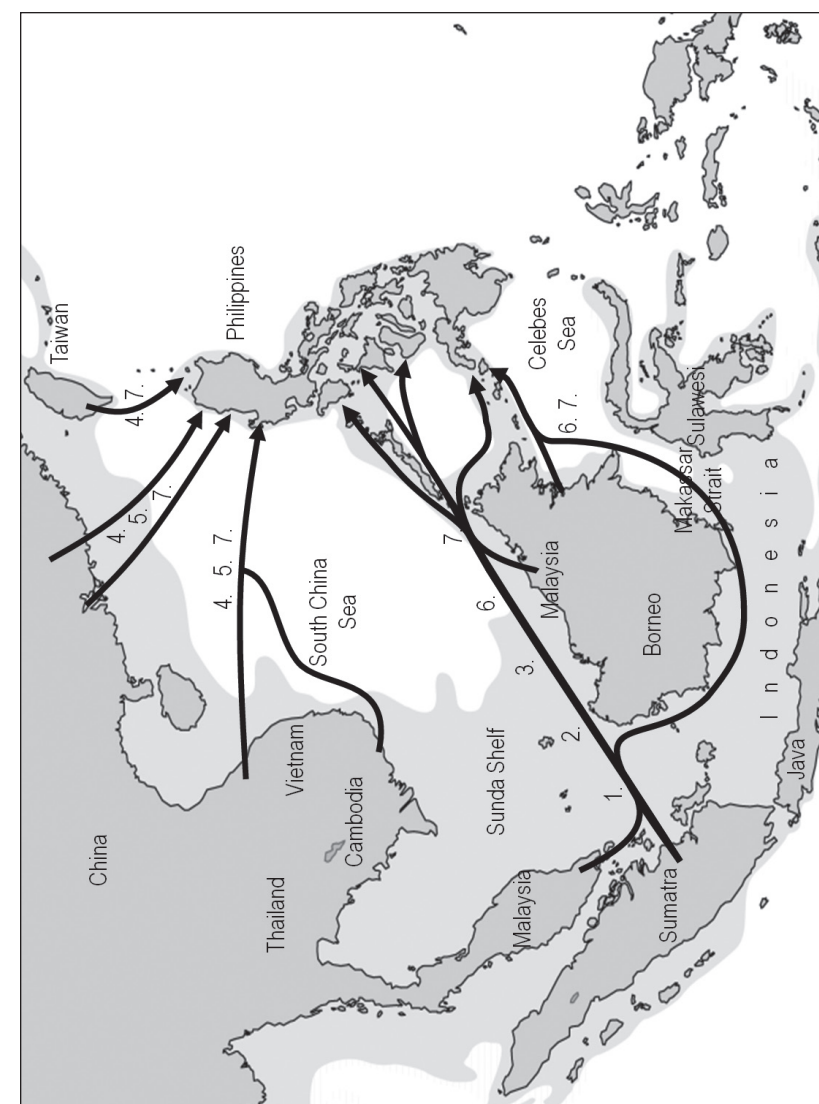


Fig. 1. Waves of Migration Theory
A graphic reinterpretation of the Waves (seven waves) of Migration Theory postulated by Henry Otley Beyer (1950, 10) with details from the theory's reevaluation by F. Landa Jocano (1989, 42).

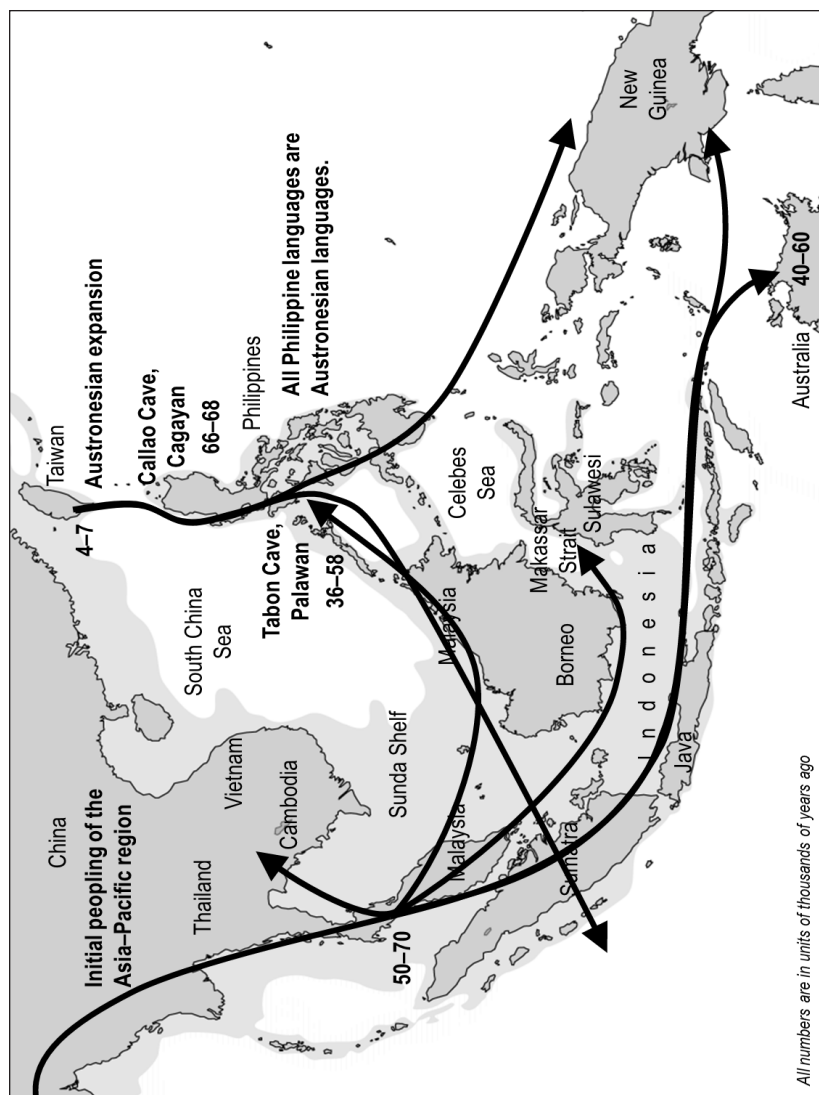


Fig. 2. Demographic events that could have influenced Philippine population genetic history and demography

are grouped into about seventeen regions that have geopolitical boundaries. Over time Filipinos have migrated to regional centers, which are now composed of inhabitants who have local (within the region) ethnolinguistic group ancestry and migrants from other regions of the Philippines. Thus, a general picture of the contemporary demographic structure of the country's population consists of specific ethnolinguistic groups, which constitute a region from its center down to its local communities and a mixture of different groups in regional centers.

The Philippine fossil records, the diversity of languages and cultures, and the diversity and demographic structure of the population truly make the Philippines relevant in the study of human diversity, population history, and human origins in the Asia-Pacific region. Studies on prehistory, history, and culture have long been the realm of archaeology, biological anthropology, and linguistics (Scott 1984; Jocano 1998; Détroit et al. 2004; Gray et al. 2009; Mijares et al. 2010). Advances in molecular biology and statistics, however, have made possible the use of genetics in the study of human demography, history, and origins. For the Philippines, Jocano (1988, 47) had even suggested that genetics was a feasible method to study prehistoric populations. Pioneering studies in the late 1970s used protein genetic markers (classical genetic markers) and focused on the genetic histories of specific Filipino groups (Omoto et al. 1978; Matsumoto et al. 1979; Omoto 1979; Horai et al. 1981; Omoto et al. 1981). Subsequent population genetic studies used DNA (deoxyribonucleic acid), commonly referred to as the blueprint or genetic code of a living (human, animal, plant, or other) organism. However, these studies only considered specific Philippine groups, associating them with demographic events and/or as part of reference data sets to study other groups in the Asia-Pacific region (Capelli et al. 2001; Hill et al. 2006; Kayser et al. 2006; Tabbada et al. 2010). To date, none have provided a view of the human genetic landscape of the Philippines using DNA.

Studying Philippine Genetics

The DNA Analysis Laboratory, Natural Sciences Research Institute, University of the Philippines, Diliman (UPD-NSRI-DAL), Quezon City, Philippines, started local studies on Philippine population genetics in 1996. The Human Population History Research Group, Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology (MPI-EVA-HPHRG) in Leipzig, Germany, started Philippine population

genetics studies in 2005. Both research groups started collaborations in 2006, with the author's acceptance into the MPI-EVA-HPHRG for his doctoral studies.

Over 1,000 DNA samples from six regional centers (RC groups) and nineteen specific ethnolinguistic groups (EL groups) were collected from 1997 to 2005 (fig. 3). All relevant permits and clearances were obtained at the institutional and community levels. The communities and individual volunteers gave their free and prior informed consent to participate in the study (The HUGO Pan-Asian SNP Consortium 2009; Delfin et al. 2011; Gunnarsdóttir et al. 2011).²

The results of our studies have been published in reputable scientific journals (The HUGO Pan-Asian SNP Consortium 2009; Tabbada et al. 2010; Delfin et al. 2011; Gunnarsdóttir et al. 2011; Maiquilla et al. 2011; Reich et al. 2011, Pugach et al. 2013; Delfin et al. 2014; Ko et al. 2014). Moreover, the results have been and are still being shared with the participating communities and other stakeholders through seminars, conferences, laboratory tours, posters, and so on (HPHRG-DAL 2014, 2015).

This paper summarizes the results of the genetic studies that our team has completed to date in order to provide a genetic perspective on the history and demography of the Philippines. Due to the highly technical nature of the field, in this paper we skip the technical data and present the results of our studies in a manner accessible to nonspecialists.

Basic Concepts and Analyses

DNA samples were analyzed in the laboratory using different DNA markers that are located in various places (genetic loci) found in diverse genetic systems across the human genome, the entirety of a person's DNA. Genetic data were analyzed using various statistical genetics methods and compared with genetic data from over a thousand individuals representing different Asia-Pacific groups. A comparison was also made with ancient DNA from an extinct, archaic human known as Denisovan, discovered in Denisova cave in Siberia (Gibbons 2011a).

The different genetic systems in the human genome consisting of autosomal DNA, Y-chromosome DNA, and mitochondrial DNA were used in our studies. Autosomal DNA or autDNA is diploid or in pairs. One half of the pair of a person's autDNA is inherited from the father and the other half is inherited from the mother. Tens to hundreds of thousands of DNA markers from different genetic loci on autDNA were used in our studies. The Y-chromosome

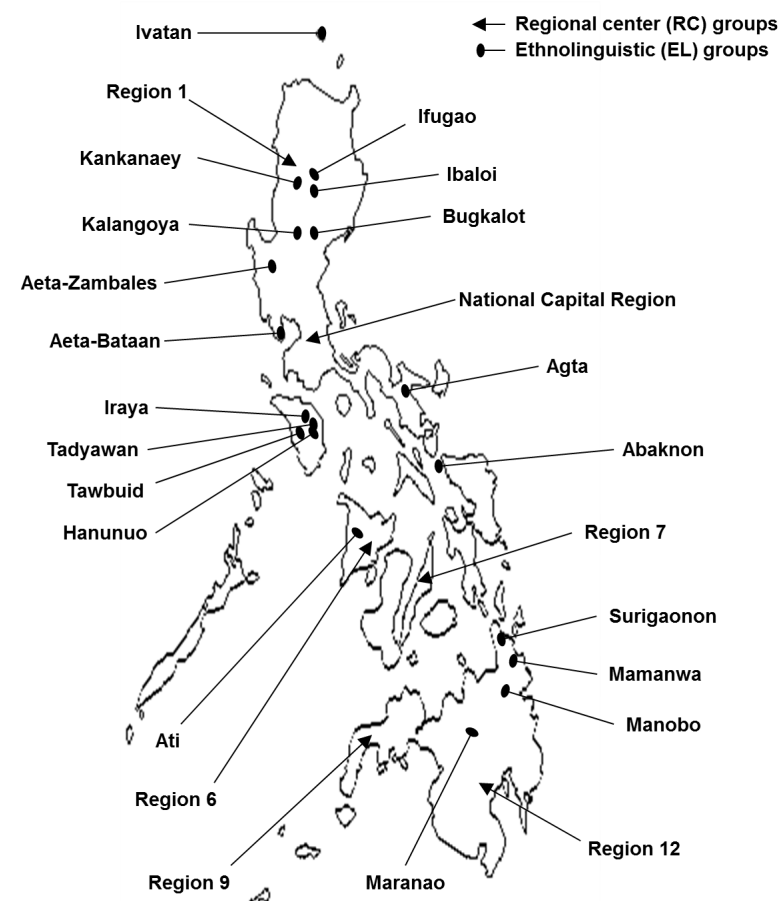


Fig. 3. Ethnolinguistic and regional center groups included in studies of Philippine genetics

or yDNA is inherited only from the father, passed along the paternal or male line. Mitochondrial DNA or mtDNA is inherited only from the mother, passed through the maternal or female line. Both yDNA and mtDNA are considered as representing a single genetic locus or place in the human genome. And since yDNA and mtDNA are each inherited only through a single line (male or female), these DNA systems are also considered haploid and are referred to as uniparental (from only one parent) DNA systems, markers, or genomes.

The analysis of genetic data is based mainly on the genetic similarities, differences, and/or variation in the DNA between individuals and among groups of individuals or populations. The term “genetic affinity” is used from this point on.

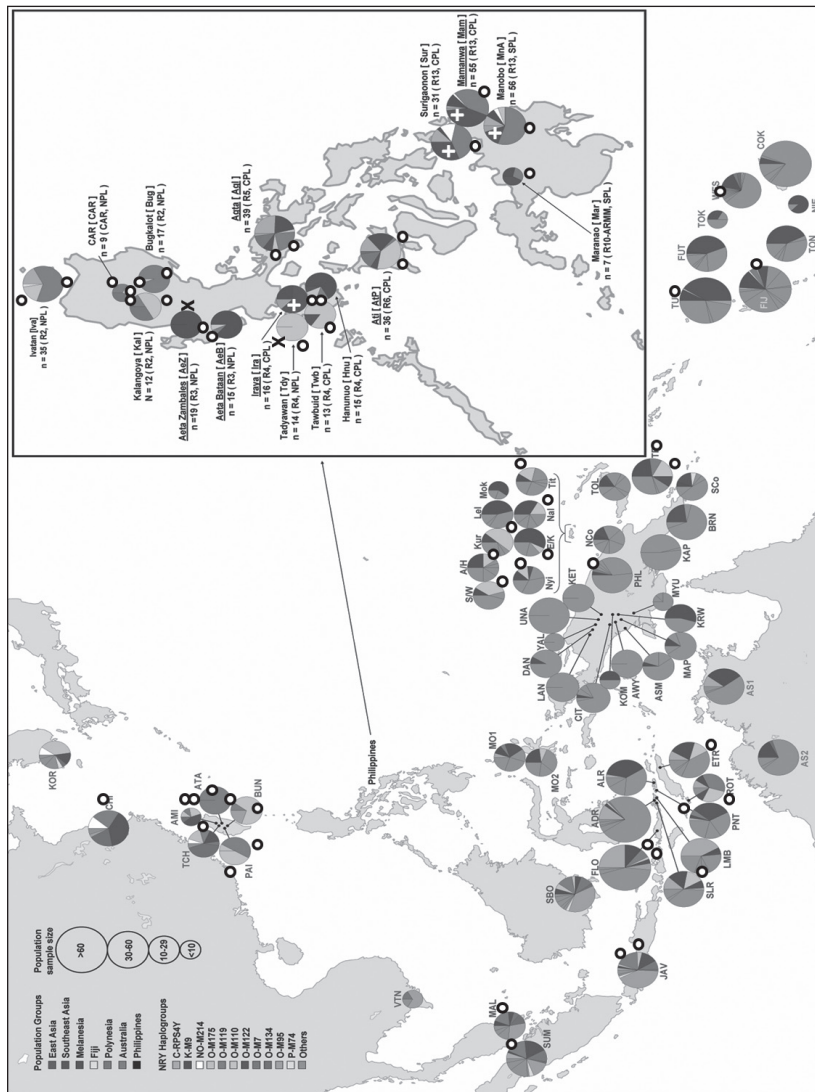


Fig. 4. An example of genetic affinities and effects of genetic drift based on yDNA. This figure is modified from fig. 1 of Delfin et al. (2011, 225). Circles track yDNA lineages from Taiwan through the Philippines, to Southeast Asia and Oceania. Philippine EL groups possibly affected by genetic drift are marked with "x." White plus sign (+) shows shared yDNA between some groups.

The analysis of genetic data also considers changes in DNA through time. Because DNA is passed down from ancestors to current generations, the DNA of people in the present can provide genetic signals of the past, stretching back hundreds of thousands of years (Jobling et al. 2004). Including ancient DNA from actual ancient fossils in the analysis can increase the resolution of genetic signals of the past (Stoneking and Krause 2011; Gibbons 2011b; Ko et al. 2014). The ages that are estimated from genetic data translate to ages of divergence or coalescence. Divergence time represents a forward-in-time perspective when populations split or diverge from a common ancestor in the past going forward into the present. Coalescent time has a backward-in-time perspective when populations in the present are tracked backward into their past until they merge or coalesce with their common ancestor.

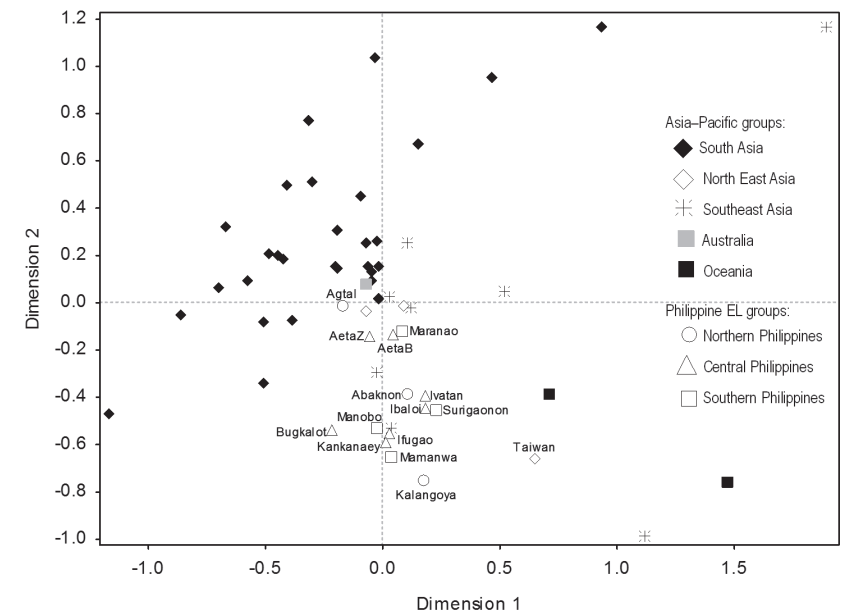


Fig. 5. Genetic affinities based on mtDNA of Philippine ethnolinguistic groups. This figure, modified from fig. 3 of Delfin et al. (2014, 235), is a plot of genetic distances between Philippine EL groups and Asia-Pacific groups. Genetic distance refers to how similar (small genetic distance) or how different (large genetic distance) groups are. The smaller the genetic distance between groups, the closer groups are to each other in the plot. The bigger the genetic distance between groups, the further away groups are from each other in the plot.

It should be noted, however, that external factors could affect the genetics of a person and a population. The relevant factors include (a) human migration, (b) genetic drift, and (c) admixture. In figure 4 (white circles), yDNA lineages found in samples from Taiwan, the Philippines, some Southeast Asian, and Oceanian groups can be due to the migration of males who possessed these yDNA from Taiwan to Southeast Asia, through the Philippines. Genetic drift is an evolutionary process whereby, due to the randomness of the inheritance of DNA through generations, not all of the DNA variation is passed on to the next generation and is lost. In figure 4 (“x” marks), genetic drift can explain why the Aeta of Zambales and the Tadyawan appear to have only one type of yDNA lineage compared with other EL groups. Admixture or gene flow is essentially the exchange of DNA as a result of mating. Apart from migration, the Mamanwa, Agusan Manobo, and Surigaonon EL groups in figure 4 have almost the same type of male lineages due to the male-driven gene flow between groups.

Other factors such as isolation (physical, geographical, linguistic, and cultural), environment (physical barriers such as mountains and bodies of water), and cultural practices (influences in mate or partner choice, postmarital residence practices, and so on) can also affect human genetics (HPHRG-DAL 2014; Jobling et al. 2004; Stoneking and Delfin 2010; Heyer et al. 2012). As a result, the genetic variation observed between individuals and among groups can provide genetic signals of such factors.

Genetic Affinities with Asia-Pacific Groups

Based on the Filipino DNA samples we collected and analyzed, Philippine groups have genetic affinities with various groups in the Asia-Pacific region. The yDNA (fig. 4) and mtDNA (fig. 5) show that Philippine EL groups have closer genetic affinities with Taiwan and Southeast Asian groups compared to other groups in the Asia-Pacific region. The same affinity was observed using autDNA (The HUGO Pan-Asian SNP Consortium 2009). Philippine RC groups also showed the same affinities (The HUGO Pan-Asian SNP Consortium 2009; Tabada et al. 2010).

Philippine EL groups such as the Aeta of Zambales, the Aeta of Bataan, the Agta of Iriga, the Iraya of Mindoro, the Ati of Panay, and the Mamanwa of Surigao are among over thirty EL groups in the Philippines that possess pygmy physical features or the pygmy phenotype (Perry and Dominy 2009, 220). They are historically and anthropologically described to have

a hunter-gatherer mode of subsistence and have been collectively referred to as “Negrito” groups (Padilla 2013, 209). However, in contrast to their collective labeling, our results (yDNA, mtDNA, and autDNA) for these six EL groups show genetic affinities with different groups in the Asia-Pacific region. Figure 5 shows that the mtDNA of the Agta of Iriga and the Aeta of Zambales and Bataan are close to Indian groups, while the Mamanwa are closer to Southeast Asian and Oceanian groups. AutDNA also show affinities with Indian groups (The HUGO Pan-Asian SNP Consortium 2009). The yDNA also shows different affinities for these EL groups (fig. 6).

For the Mamanwa EL group, their yDNA show similarities with Melanesian groups, but not with Australian groups. However, autDNA show that there are genetic affinities between the Mamanwa and Australian groups (Pugach et al. 2013, 1803).

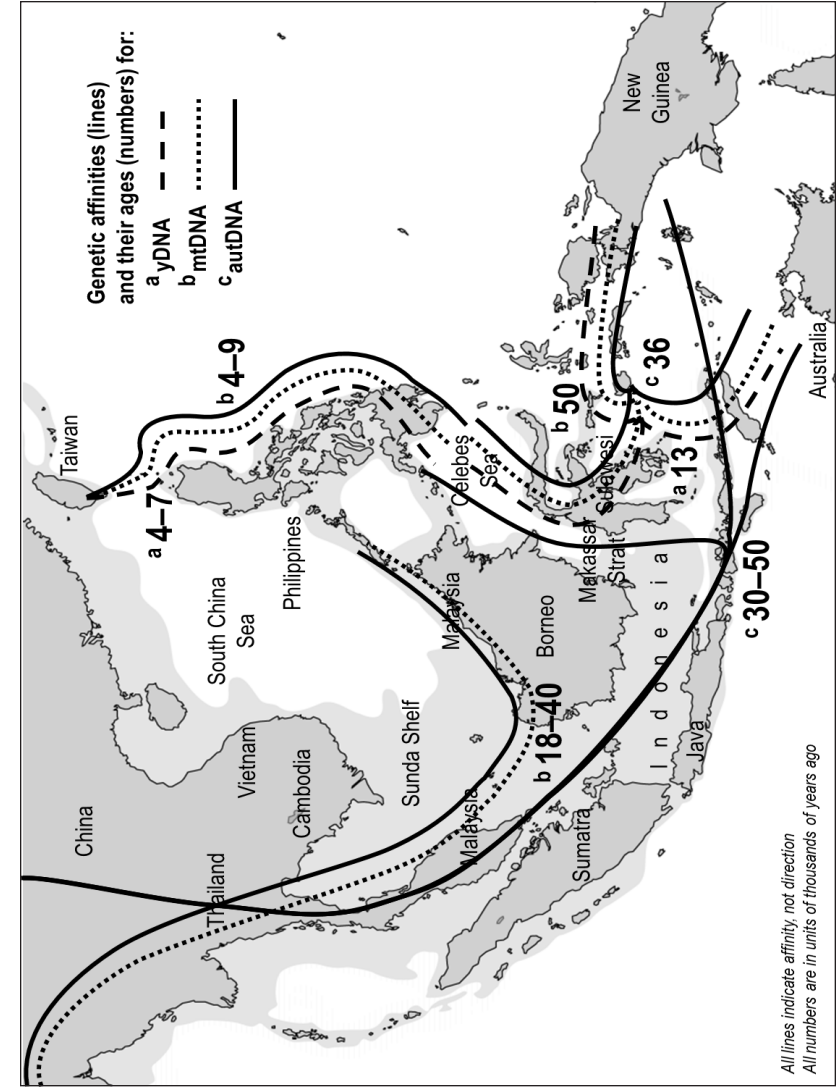
Due to technical limitations, Mamanwa and Agusan Manobo DNA were the only Philippine EL groups compared to ancient Denisovan DNA. Genetic affinities were observed between ancient Denisovan DNA and Asia-Pacific groups, in particular Australian and Melanesian groups, including the Mamanwa (Reich et al. 2011, 516).

The preceding paragraphs described some of the genetic affinities observed. Figure 7 summarizes all the genetic affinities observed in our studies so far. In general, Philippine groups have genetic affinities with groups from South Asia to Australia, even with archaic humans such as Denisovan; however, Philippine groups appear to have closer affinities with groups from Southeast Asia and Taiwan than with other groups from South Asia to Australia (The HUGO Pan-Asian SNP Consortium 2009; Tabada et al. 2010; Delfin et al. 2011; Pugach et al. 2013; Delfin et al. 2014; Ko et al. 2014). As previously discussed, genetics can be affected by external factors. Different affinities for each EL group could suggest different group contacts and admixture in the Asia-Pacific region. Affinities to the same Asia-Pacific group may suggest similar group contact. Alternatively, an Asia-Pacific group may have had contact and admixed with one EL group that then admixed with another EL group, resulting in shared affinity with the Asia-Pacific group. This pattern was observed in the admixture of Denisovan, Mamanwa, and Manobo. Both Mamanwa and Manobo show signals of admixture with Denisovan, but further analysis has shown the Manobo acquired the Denisovan admixture signal by admixture with the Mamanwa (Reich et al. 2011, 516). Male lineages (yDNA) and female

male lineage of
L groups

age:
0-19,999 years ago
20-34,387 years ago

Fig. 6. Genetic affinities based on yDNA of Philippine ethnolinguistic groups with the pygmy phenotype. *This figure is modified from fig. 3, K-MG of Delfin et al. (2011, 228). Labeled are Philippine EL groups and the Asia-Pacific groups with which they have genetic affinity.*



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lineages (mtDNA) could also have independent histories resulting in different genetic affinities.

All these suggest a complex genetic history. The Philippines is a diverse and multicultural country, with influences from different parts of the Asia-Pacific region. The genetic affinities between Philippine groups and Asia-Pacific groups provide a genetic perspective to the associations that Philippine groups have had with other groups in the region. It further shows a complex history of where the ancestors of Filipinos and Asia-Pacific groups might have exchanged or shared cultures and DNA as well.

Genetic Signals of Demographic Events in the Past

The initial peopling of the Asia-Pacific region and the Austronesian expansion (fig. 2) are two major demographic events that have influenced Philippine history.

In terms of the initial peopling of the Asia-Pacific region, the genetic affinities that Philippine groups have with other Asia-Pacific groups and their respective ages of about 40,000–50,000 ya (fig. 7), which overlap with the ages of fossil records, can be interpreted as genetic signals of the initial peopling of the Philippines and the Asia-Pacific region. These observations also suggest that the ancestor of Filipinos could be part of the initial settlement of ancient humans who migrated via a southern coastal route through South Asia into Southeast Asia. Notable is the possible gene flow between the ancestors of the Mamanwa EL group and the archaic Denisovan group that could have happened prior to the initial settlement of the Philippines (Reich et al. 2011, 516).

For the Austronesian expansion, the genetic affinities and their respective ages that Taiwan, Philippine, and other Asia-Pacific groups have (figs. 4 and 6) are said to be signatures of the Austronesian expansion (Kayser et al. 2008; The HUGO Pan-Asian SNP Consortium 2009; Ko et al. 2014). That is, the Austronesian speakers who migrated from Taiwan through the Philippines and into the Asia-Pacific region are of these lineages. The yDNA, mtDNA, and autDNA affinities that Philippine groups have with Taiwan and other Southeast Asian and Pacific groups (fig. 7; about 4,000–9,000 ya) suggest that Philippine groups, apart from their languages, possess genetic signals of the Austronesian expansion. These genetic signals also suggest that Austronesian speakers did not only share their language but also their DNA.

Genetic affinities whose timelines are more recent than the initial peopling, but older than the Austronesian expansion of about 13,000–40,000

ya (fig. 7), can indicate human migrations and gene flow apart from those that occurred during the major demographic events. Overall, a timeline of genetic similarities ranging from about 5,000–50,000 ya suggests an ancient genetic ancestry for the Philippine population.

A Complex Population Genetic Structure

Figure 8 illustrates a general trend of genetic closeness and distance (or difference) between Philippine RC and EL groups. This general trend, which is consistent across yDNA, mtDNA, and autDNA, describes a Philippine population genetic structure in which RC groups are genetically closer to each other and to some EL groups, while EL groups in general are genetically distant from each other (HPHRG-DAL 2014, 2015).

Notable differences are seen among the Tadyawan, Tawbuid, Hanunuo, and Iraya EL groups that, despite all of them being found on the island of Mindoro, are genetically distant from one another compared with the closeness among RC groups. Consistent with their affinities with different groups in the Asia-Pacific region, the six EL groups collectively called “Negritos” are genetically distant. Comparing for example the yDNA (fig. 9) among the six EL groups, including other EL groups that are of the same male lineage, consistently shows in general that they are genetically distant from each other. Notably the differences or divergence shown by yDNA are consistent with the contemporary geographic situation of these EL groups that are isolated from each other. Moreover, this divergence or separation is estimated to have occurred in the Philippines’s prehistoric past of about 10,000 ya. An exception is the Iraya, which appears to be close to the Mamanwa, possibly because of shared yDNA (fig. 4, white plus sign) and autDNA (fig. 8).

The genetic results appear to be consistent with the oral tradition that describes the relationship between the Aeta of Bataan and those of Zambales. The narratives state that the Aeta of Bataan were once part of the Aeta group in Zambales and that a male and female couple left the Zambales group to start their own group in Bataan (Tebtebba Foundation 2008, 13). This oral tradition sheds light to the data that genetically the Aeta of Bataan and those of Zambales, although not completely overlapping, are close to each other (figs. 7, 8, and 9). The yDNA (fig. 8) may also have captured the image described in oral tradition such that there is a cluster of Aeta of Bataan and Zambales in one network branch, while there is a single Aeta of Bataan yDNA that is separated from the Bataan-Zambales cluster.

This plot is similar to fig.



Fig. 9. Philippine
ethnolinguistic (EL)
groups with the pygmy
phenotype and other EL
groups under one yDNA
lineage



Overall our studies illustrate a complex Philippine population genetic structure composed of a homogeneous pool of RC groups and a heterogeneous pool of EL groups. This complex structure demonstrates the diversity and genetic variation in the Philippine population. Consistent with population genetic studies of other countries and populations (Jobling et al. 2004, 278), genetic variation is greater within Philippine groups (68–90 percent) and lesser between Philippine groups (9–31 percent) (Delfin et al. 2011, 2014). These percentages support the argument against the use of the term race or *lahi* to refer to Filipinos (*lahing Pilipino*) or to refer to ancestry from a particular Philippine group (lahing Ilokano, Bikolano, and so on), to foreign groups (lahing Amerikano, Italiano, and so on), or to any racial grouping in general.

An Alternative Origins Story

Given the contemporary size and diversity of the Philippine population, the population coverage of our studies is basically a scratch on the surface of this population. However, the genetic overview that our studies provide gives insights into a Philippine genetic history that is complex and with possible influences of factors such as genetic drift, migrations, and admixture from outside the Philippines and between Philippine groups. As shown on figures 1, 2, and 7, the origins of the Filipino people may no longer be as straightforward as it was previously assumed, starting from an empty archipelago that received a successive influx of different groups of people. A multidisciplinary evaluation that brings together current anthropological and genetic evidence can provide another perspective to the story of origins.

This alternative origins story of the Filipino people (fig. 7) starts with the initial peopling of the Asia-Pacific region about 50,000–70,000 ya by the human ancestors of Asia-Pacific groups (O'Connell et al. 2004; Barker et al. 2007). This initial occupation included ancestors whose presence in the Philippine archipelago occurred as early as 47,000–67,000 ya (Détroit et al. 2004; Mijares et al. 2010) and supported by genetically derived age estimates of as old as about 50,000 ya (Delfin et al. 2011; Gunnarsdóttir et al. 2011; Reich et al. 2011; Delfin et al. 2014). These early ancestors could have included Mamanwa ancestors who had admixed with Denisovans (Reich et al. 2011). In the initial occupation, ancient humans traversed a southern coastal route through South Asia into Southeast Asia and the Pacific (Jobling et al. 2004, 283). The genetic affinities that Philippine groups share with

groups from South Asia to Australia suggest that the ancestors of Philippine populations were part of this southern coastal migration (The HUGO Pan-Asian SNP Consortium 2009; Delfin et al. 2011; Reich et al. 2011; Pugach et al. 2013; Delfin et al. 2014).

In the initial settlement of the region, human groups would have found their first niches. Depending on the environment, the chosen niches could have isolated groups from each other. Alternatively the early humans in the region did not only settle but also moved around after their initial settlement. In the case of Philippine ancestors, the Philippine archipelago would have provided an environment that could have isolated groups from one another, yet with the possibilities for movement by land or water (Scott 1984; Jocano 1998). The different genetic affinities that Philippine groups have with Asia-Pacific groups and their respective age estimates of 13,000–50,000 ya (Delfin et al. 2011; Pugach et al. 2013; Delfin et al. 2014) suggest that Filipino ancestors had contact with other groups in the region after initial settlement. In general, apart from human migration during the initial settlement, subsequent migrations that led to contact between groups at different times after initial settlement could have occurred.

After the initial peopling, the Austronesian expansion followed as the most important demographic event that had a major impact on Philippine ancestry. Originating from Taiwan 4,000–7,000 ya, Austronesian speakers migrated through the Philippines (Bellwood and Dizon 2005; Gray et al. 2009). They had contact with Philippine ancestors, which led to the sharing of cultures and languages (Bellwood 1984; Bellwood and Dizon 2005). Presumably some Philippine EL groups spoke non-Austronesian languages, which were subsequently replaced by Austronesian languages (Reid 1994) such that all contemporary Philippine languages are of the Austronesian language family (Lewis et al. 2015).

The genetic affinities that Philippine groups have with Taiwan and other groups in Southeast Asia and their estimated ages of about 4,000–9,000 ya provide genetic signatures of the Austronesian expansion (The HUGO Pan-Asian SNP Consortium 2009; Tabbada et al. 2010; Delfin et al. 2011; Delfin et al. 2014; Ko et al. 2014). The genetic evidence also suggests that the Austronesians did not only share their culture and replaced the local languages, but they also admixed with the Philippine ancestors. The genetic perspective incorporated so far in this origins story is localized in the Late Pleistocene and Early Holocene time periods of Philippine prehistory. This

timeline overlaps with the second to fourth migration waves proposed by Beyer (1950) and the early parts of a Formative prehistoric culture phase discussed by Jocano (1998). The subsequent migration waves and phases of prehistoric culture development would have entailed more migrations, contact, culture exchange, and admixtures.

The Complexity of the Philippine Population Structure

A genetic perspective of the Philippine population structure illustrates a complex demography with a heterogeneous pool of EL groups and a homogeneous pool of RC groups. The center of administrative regions had seen the concentration of government offices, economic activities, educational institutions, and services. This centralization at the regional level was accompanied by migration and settlement, resulting in the formation of regional center populations. Eventually internal migration would no longer be limited to short-distance movers, who moved between cities within regions, but also involved long-distance movers, who migrated between administrative regions. The internal long-distance migrations resulted in intermarriages among different Philippine population groups and genetic admixtures, helping explain the homogeneity of the genetic pool of RC groups.

In the case of the heterogeneous pool of Philippine EL groups, their genetic differences could be surmised from the alternative origins story previously described. The ancestors of Philippine EL groups could have found their respective niches in the archipelago early in Philippine prehistory, occupying these territories and passing them on to later generations such that contemporary EL groups consider these territories as their ancestral lands.

Figure 9 shows that, while some EL groups belong to the same male lineage, they diverged from their common ancestor more than 10,000 ya. Figure 9 also shows that the positions of the EL groups in the network roughly correspond to their current geographical positions. The time period in this genetics example is in the Pleistocene, a time in Philippine prehistory without written records. Differences in Philippine EL groups could also have resulted from group isolation, which might have been due to factors such as geography, culture, language, and socioeconomics. In terms of geography, EL group settlements could range from mountainous, landlocked environments to waterlocked islands. Each EL group's language is basically unintelligible to the other. Cultural factors such as marriage customs would have prevented

admixture between groups. For instance, despite geographical proximity on the island of Mindoro, the genetic difference between the Hanunuo, Tadyawan, Tawbuid, and Iraya could be due to language barriers and the custom of marrying only within the same EL group (Lebar 1975).

Although the term race (*lahi*) is commonly used, the concept of race is a social construct and has no biological or genetic basis. In biology race is equivalent to the species or subspecies level or classification. All humans all over the world, including Filipinos whether belonging to RC groups or EL groups, are of the same species, *Homo sapiens*, or subspecies, *Homo sapiens sapiens*. Genetic variation is greater within groups than between groups that are thought to form a race. The percentages of genetic variation estimated in our studies (Delfin et al. 2011, 2014) are consistent with previous research (Jobling et al. 2004) and thus provide support against racial grouping.

In terms of Philippine groups, the collective naming of EL groups that have pygmy features and their collective distinction from other EL groups without pygmy features is perpetuated without genetic basis, given differences between these EL groups. Similarly the collective labeling of EL groups on the island of Mindoro as “Mangyan,” which can be pejorative, also has no genetic basis (Delfin et al. 2011, 2014).

DNA Changes over Time

DNA can change over time. These changes may be neutral and have no effect on the organism while some changes can have negative effects (such as diseases) or positive effects (such as resistance to disease). Studies have shown that throughout human history (and prehistory), changes in DNA resulted in physical traits that allowed humans to, among others, adapt to environmental conditions and resist infection (Vitti et al. 2012; Karlsson et al. 2014). Given a genetic ancestry that could be as old as 5,000–55,000 ya and genetic affinities that could have resulted from admixtures, there could have been changes in the DNA of Philippine groups over a long period of time.

For instance, the admixture of archaic humans (Neanderthals and Denisovan) with the ancestors of contemporary humans showed that archaic humans could have contributed genes relevant to adaptation such as those that affect skin and hair or genes related to diseases like diabetes or lupus (Sankararaman et al. 2014, 354). The admixture of Denisovan with the ancestors of the Mamanwa (Reich et al. 2011, 516) could have resulted

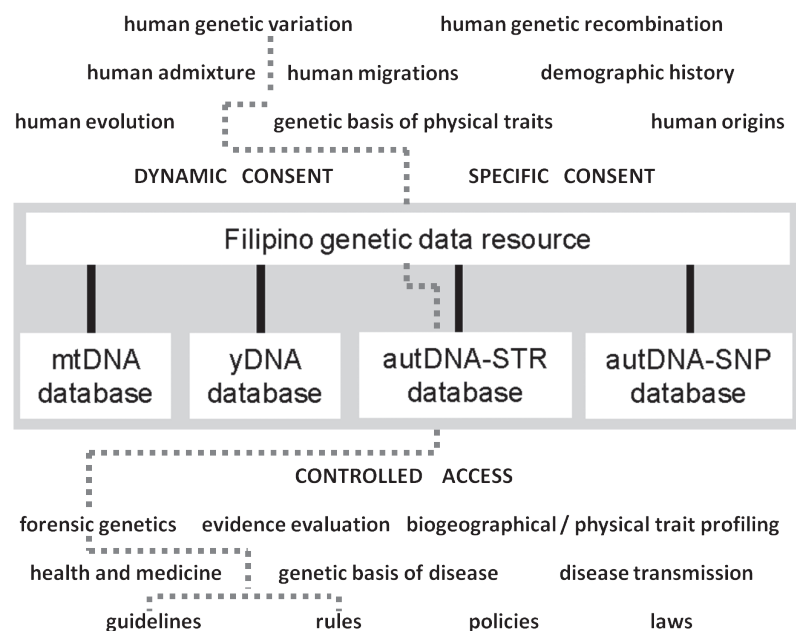


Fig. 10. A Philippine genetic data resource for various studies and applications

The data resource is composed of databases of different genetic information: mtDNA, yDNA, autDNA-STR (Short Tandem Repeats),³ autDNA-SNP (Single Nucleotide Polymorphism),³ useful in basic research (terms above the data resource) and applied research (terms below the data resource). Dotted lines refer to research application.

in contemporary Mamanwa possessing genes similar in functions. These possible changes in DNA are yet to be studied in the Mamanwa and in Philippine populations overall. The characterization of these changes and the effects they could have produced (whether neutral, positive, or negative) would provide significant insights into Philippine demography, history, and possibly health and medicine.

A complex population genetic structure (fig. 8) has several implications. In the use of DNA information for the evaluation of DNA evidence, a single database for regional populations may be sufficient because RC groups make up a homogeneous pool (Tabbada et al. 2002; Maiquilla et al. 2011). In contrast the genetic diversity of EL groups necessitates the inclusion of an appropriate correction factor when using the Philippine database in order to account for the population substructure. Alternatively specific databases

for EL groups may be used for the statistical analysis of DNA evidence. In terms of history, evolution, and medicine, the homogeneity of the RC groups could mean that they share evolutionary histories in which changes in DNA and the consequences of these changes for medicine and human health would be the same across all RC groups. In contrast different EL groups could have independent demographic and/or evolutionary histories and the consequences of the changes in DNA that accompany these histories would be group specific.

Future Population Genetics Research

The story of origins, history, and demography of the Philippines that is told here ends for the moment. Although a mere scratch on the surface, our research has revealed a complex genetic history and demography for the Philippine population with the possible influence of various evolutionary, historical, and demographic factors. To continue the story, to address the succeeding stages of the origins of the Filipino, the different histories and demography of Philippine groups will require genetics to have greater representation of Philippine groups and greater coverage of the Filipino genome. To this end, succeeding studies will need to adopt a comprehensive study protocol for research on Philippine population genetic/genomic variation.

The goal is to have a Philippine genetic data resource (fig. 10) that will be useful for both basic research (such as genetic variation, recombination, and evolution) and applied research (such as forensics and medicine). Through the relevant ethics instruments (consent and access), the genetic data resource will protect and empower the Filipino people. The use and application of this genetic data resource can lead to relevant instruments (such as policies and guidelines) that can serve Philippine society. For example (fig. 10, dotted line from human genetic variation to guidelines and rules), local efforts of the DNA Analysis Laboratory have led to the establishment of a Philippine genetic database of standard forensic DNA markers such as Short Tandem Repeat markers or STRs³ and the incorporation of forensic genetics in the local legal-justice system (De Ungria et al. 2002; De Ungria et al. 2005). These efforts have eventuated in a formal ruling on DNA evidence (Philippine Judicial Academy 2007, 5). Overall, an expanded Philippine genetic data resource will be most useful in both local and international human genetics research and applications.

Abbreviations Used

autDNA	autosomal DNA
EL	ethnolinguistic
MPI-EVA-HPHRG	Human Population History Research Group, Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology
mtDNA	mitochondrial DNA
RC	regional center
STR	short tandem repeat
SNP	single nucleotide polymorphism
UPD-NSRI-DAL	DNA Analysis Laboratory, Natural Sciences Research Institute, University of the Philippines Diliman
ya	years ago
yDNA	Y-chromosome DNA

Notes

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- 1 “Asia-Pacific” refers to the geographic region that includes South Asia (India), northern East Asia (China, Japan, Taiwan), Southeast Asia (from Thailand to Indonesia to the Philippines), Australia, and Oceania (Melanesia and Polynesia). This area is relevant in the study of human

origins as it includes the route (South Asia) that ancient humans took in entering the region, the regions that were settled the earliest (Southeast Asia and Australia), as well as regions that were affected by subsequent human dispersals like the Austronesian expansion (Southeast Asia, Oceania).

- 2 Most of our DNA sample collections were conducted from 1997 to 2005 and thus predate the recent ethics policies and regulations. The papers cited in this paragraph are those for which the relevant ethics policies and regulations are applied.
- 3 STRs are DNA markers found on autDNA and are routinely used in forensic genetics. SNPs are the most abundant type of DNA markers found on autDNA, across the entire human genome.

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