Available online at www.sciencedirect.com
ScienceDirect
Current Opinion in Genetics & Development

Recent developments in the genetic history of East Asia and Oceania

Ana T Duggan and Mark Stoneking

Recent developments in our understanding of the genetic history of Asia and Oceania have been driven by technological advances. Specifically, our understanding of the past has been augmented by: genome sequences from ancient hominins and ancient modern humans; more comprehensive studies of existing populations (e.g., complete mtDNA genome sequences and genome-wide data) and the development of new statistics and analytical methods to interpret the abundance of new data. We review some of the new discoveries since we entered the age of archaic and modern genomics and how they have changed our understanding of the settlement and subsequent population dynamics in Asia and the Pacific.

Addresses
Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Deutscher Platz 6, D04103 Leipzig, Germany

Corresponding author: Stoneking, Mark (stoneking@eva.mpg.de)

Current Opinion in Genetics & Development 2014, **29**:9–14

This review comes from a themed issue on **Genetics of human evolution**

Edited by **Aida M Andrés** and **Katja Nowick**

For a complete overview see the Issue and the Editorial

Available online 23rd August 2014

http://dx.doi.org/10.1016/j.gde.2014.06.010

Introduction

East Asia and Oceania, defined here as that part of Eurasia bordered by the Ural Mountains to the west and the Himalayan Plateau to the south, and extending eastwards through island southeast Asia, Australia, New Guinea, and the farthest reaches of Polynesia (Figure 1), is a vast area that encompasses a wide range of environments, peoples, cultures, and languages. Home to some of the earliest fossil evidence for modern humans outside of Africa [1–3], east Asia and Oceania witnessed both the earliest dispersal of modern humans from Africa more than 40,000 years ago (probably via a southern route, as discussed in more detail below) as well as the most recent, involving the initial colonization of Pacific islands that reached such far-flung Polynesian islands as Hawai'i, New Zealand, and Easter Island by less than 1000 years ago [4]. Genetic studies have tried to estimate the impact and contribution of these (and other) dispersals to the genetic structure of contemporary populations, and in this review we will focus on what we view as the most significant new developments since previous reviews on the genetic history of east Asia and Oceania [5,6]. In particular, there are four notable advances that have already contributed substantially, and promise to contribute much more, to our knowledge of the genetic history of humans in general and human populations in this part of the world in particular: first, insights from archaic human ancestry in modern human populations; second, unbiased samples of complete mtDNA (and, increasingly, partial Y chromosome) sequences; third, more genome-wide data, both from SNP arrays and from genome sequencing; and fourth, substantial advances in how to make use of all of these new data to infer demographic parameters of interest (including identifying and dating admixture events), using model-based and other approaches.

Archaic human ancestry

Thanks to new advances in extracting DNA from fossils and preparing sequencing libraries from ancient DNA, the original, low-coverage 'draft' genome sequences of a Neandertal [7] and a Denisovan [8] that were riddled with sequencing errors have been supplanted with high coverage genome sequences [9,10•] that, by any standard, are of the same quality as those generated from living people. And although Neandertals and Denisovans are extinct, these genome sequences show that their ancestry lives on in people today. All non-Africans carry a signature of Neandertal ancestry of about 1.9–4% that is slightly but significantly higher in east Asians than in Europeans [9,11]. It thus appears that after modern humans dispersed from Africa around 60,000 years ago, a single ancestral non-African population of modern humans interbred with Neandertals. All non-African populations today are descended from this ancestral population and thus carry this Neandertal ancestry, with some additional Neandertal interbreeding that occurred in the ancestry of east Asians.

Fossil remains of the archaic humans known as Denisovans are only known till date from Denisova Cave in southern Siberia, yet the modern human populations with the highest amount of Denisovan ancestry (about 4–6%) are found in New Guinea and Australia, some 7000 km away [8,9,12••]. Based on a comprehensive study of genome-wide SNP data, Denisovan ancestry was found to be restricted to Australians, New Guineans, the Mamanwa — an aboriginal 'Negrito' group from the Philippines — and populations known to share recent

Figure 1

Map of East Asia and Oceania, with ages of the earliest indications of human colonization indicated for selected sites. The red star denotes the location of Denisova Cave, while the dotted black line indicates the boundary between Near and Remote Oceania.

ancestry with these, such as those from eastern Indonesia, Fiji, and Polynesia [12••]. Moreover, the model that best fits the genome-wide data supports the 'early Southern route' hypothesis, which holds that the earliest dispersal of modern humans was via a southern route, and went as far as Australia and New Guinea [13,14]. This early Southern route scenario also receives strong support from a different analysis of genome-wide SNP data [15•] which used an approximate Bayesian computation (ABC) framework (including a novel approach to account for the ascertainment bias in SNP array data) to test various models of population history and estimate associated demographic parameters, as well as from a recent study that used both genome-wide SNP and craniometric data to analyse the spatiotemporal predictions of various models [16].

However, the picture of Denisovan ancestry in modern human populations has recently become more complicated, as new analyses based on the high-coverage archaic genome sequences [9,10•] find a barely-detectable signal of Denisovan ancestry of about 0.2% in all east Asians analysed (specifically, Han and Dai groups from China). Determining the extent of this low-level admixture across east Asia, and whether this east Asian Denisovan ancestry represents the same, or a different, admixture event as that detected in the Mamanwa/Australians/New Guineans should shed light on the timing and location(s) of the admixture between Denisovans and modern humans.

Austronesian origins and expansion

The Austronesian language family is one of the most widespread in the world, extending from Taiwan through