



The genetic makings of South Asia

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South Asia is home for more than a billion people culturally structured into innumerable groups practicing different levels of endogamy. Linguistically South Asia is broadly characterized by four major language families which has served as access way for disentangling the genetic makings of South Asia. In this review we shall give brief account on the recent developments in the field. Advances are made in two fronts simultaneously. Whole genome characterisation of many extant South Asians paint the picture of the genetic diversity and its implications to health-care. On the other hand ancient DNA studies, which are finally reaching South Asia, provide new incites to the demographic history of the subcontinent. Before the spread of agriculture, South Asia was likely inhabited by hunter-gatherer groups deriving much of their ancestry from a population that split from the rest of humanity soon after expanding from Africa. Early Iranian agriculturalists mixing with these local hunter-gatherers probably formed the population that flourished during the blossoming of the Indus Valley Civilisation. Further admixture with the still persisting HG groups and population(s) from the Eurasian Steppe, formed the two ancestral populations (ANI and ASI), the north-south mixing pattern of whom is known today as the 'Indian Cline'. Studies on natural selection in South Asia have so far revealed strong signals of sweeps that are shared with West Eurasians. Future studies will have to fully unlock the aDNA promise for South Asia.

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Current Opinion in Genetics & Development 2018, 53:128–133

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

Edited by **Lluís Quintana-Murci** and **Brenna Henn**

<https://doi.org/10.1016/j.gde.2018.09.003>

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Out-of-Africa and into South Asia

Recent studies harnessing the power of analysing complete genomes from many populations worldwide have established that most if not all humans outside Africa descend from the main Out-of-Africa (OoA) expansion

some 50 thousand years ago (kya) [1,2,3,4^{*}]. Given the documented admixture of all non-Africans with Neanderthals and ample archaeological evidence of Middle Palaeolithic hominin presence in South Asia well before the main OoA [5], it remains of interest to explore the vast and structured genetic landscape of South Asia for vestiges of potential additional archaic admixture events. Indeed, it was recently suggested that South and South-east Asians do carry a small proportion of genetic ancestry from an unknown extinct hominin [4^{*}]. While, these conclusions were not replicated in different datasets [6], the discussion is still open [7]. In a different bid, using a haplotype sharing pattern-based approach, Pagani *et al.* [3] claimed that at least 2% of the genomes of modern Papuans originate in an earlier OoA (xOoA) that split from the rest of Africans around 120 kya. The possibility of an earlier OoA is circumstantially backed up by evidence for admixture of modern humans into Neanderthals long before OoA [8,9] and the growing archaeological body of evidence of modern human presence in Arabia [10] and East and Southeast Asia well before 60 kya [11^{*}]. While the parallel studies [1,2] did not find evidence for genomic survival of xOoA in the Papuans, they noted that a small percentage of such ancestry may go unnoticed by the methods used in these studies. If xOoA is true, South Asia would be a prime place for vestiges of it to have survived. So far such evidence has not been presented.

Formation of the current South Asian populace – the great mix

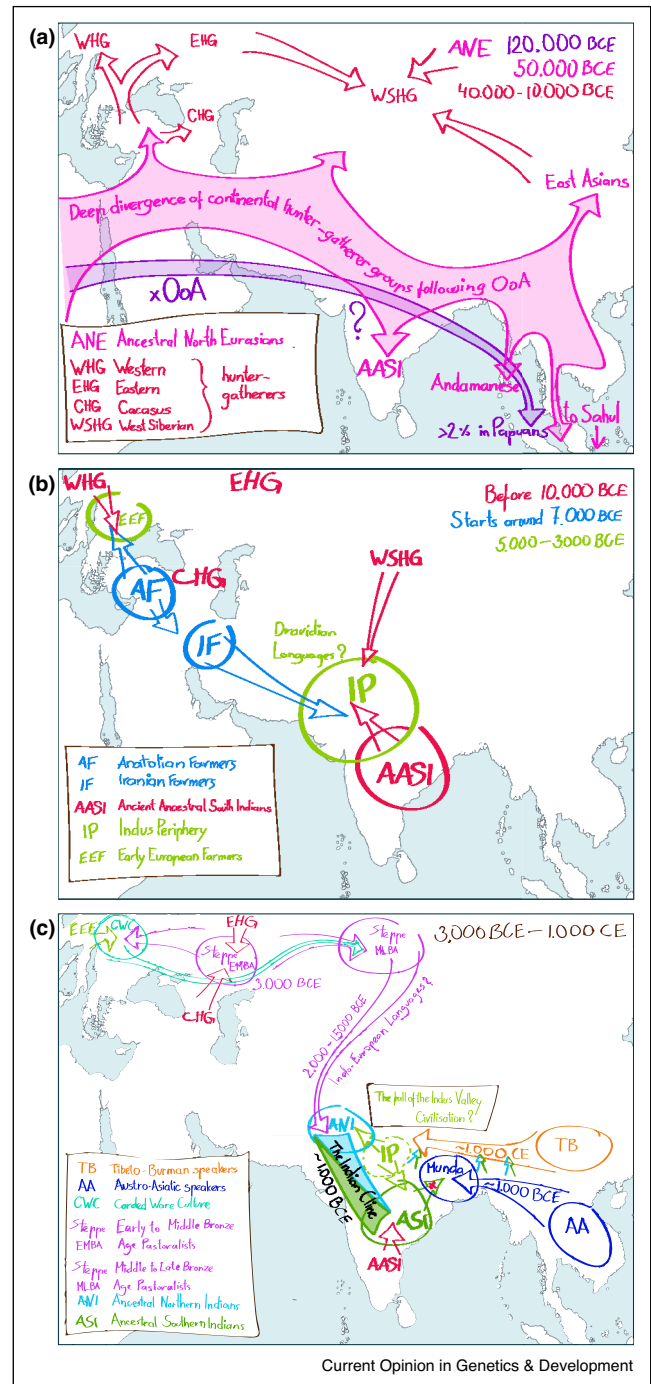
The rich literature on human mitochondrial DNA and Y chromosome phylogenetics/phylogeography in South Asia [12–18] is largely in agreement highlighting the first, presence of deep rooting autochthonous genetic component and second, shared genetic component with West Eurasians. This western component decreases in frequency from northwest (40%) toward south and east (<10%) and is more prevalent in high-caste groups (e.g. Brahmins). While it was evident that this genetic sharing originated likely in different time horizons, it was difficult to examine/test-specific admixture scenarios.

In 2009 the seminal study by Reich *et al.* [19] turned the page by conducting the first genome-wide survey in South Asia. In parallel with creating a new toolkit for population genetics analyses [20], this study revealed on the genomic level that most South Asian populations can be described as mixtures of different proportions of two ancestral populations - ancestral Northern and Southern Indians - ANI and ASI - of which the former is genetically

closer to West Eurasian genetic variation than to ASI. ASI in turn is as distant from West Eurasian branch than from East Asian (or the two latter from each other). The ANI component has a familiar geographical spread pattern in South Asia decreasing from the northwest - named in Reich *et al.* the ‘Indian Cline’. Metspalu *et al.* [21] extended this work by adding data on another 30 Indian populations. While confirming the existence of the two components, an attempt was made to date them with an *ad hoc* approach relying on haplotype diversity. Proper dating of the ANI-ASI admixture event, measuring the decay of linkage disequilibrium generated by the admixture [22], was performed by Moorjani *et al.* [23] which resulted in mixture dates ranging from about 1900 to 4200 years ago (2200 BCE–100 CE). Importantly, after this period of mixing Indian populations apparently shifted toward endogamy [23,24].

The myriad of recent ground-breaking ancient DNA studies have reshaped our understanding of the demographic history particularly of West Eurasia. Unfortunately the climatic conditions in South Asia make it difficult for human (skeletal) remains, let alone DNA in the remains, to survive for thousands (even hundreds) of years. Nevertheless, after it was shown that the spread of agriculture in Europe was a result of the demic diffusion of early Anatolian farmers, it was discovered that the spread of agriculture to South Asia was mediated by a genetically completely different farmer population in the Zagros mountains in contemporary Iran (IF) [25,26,27]. The ANI-ASI cline itself was interpreted as a mixture of three components genetically related to Iranian agriculturalists, Onge and Early and Middle Bronze Age Steppe populations (Steppe_EMBA) [25]. The first ever autosomal aDNA from South Asia comes from Northern Pakistan (Swat Valley, early Iron Age) [28]. This study presented altogether 362 aDNA samples from the broad South and Central Asia and contributes substantially to our understanding of the evolutionary past of South and Central Asia. The study redefines the three genetic strata that form the basis of the Indian Cline. The *Indus Periphery (IP)* component is composed of (varying proportions of): first, *IP*, second, Ancient Ancestral South Asians (*AASI*), which represents an ancient branch of human genetic variation in Asia arising from a population split contemporaneous with the splits of East Asian, Onge and Australian Aboriginal ancestors (in accordance with [4,29]) and third, West_Siberian Hunter gatherers (*WS_HG*). The authors argue that *IP* could have formed the genetic base of the Indus Valley Civilization (IVC). Upon the collapse of the IVC *IP* contributes to the formation of both *ASI* and *ANI*. *ASI* is formed as *IP* admixes further with *AASI*. *ANI* in turn forms when *IP* admixes with the incoming Middle and Late Bronze Age Steppe (*Steppe_MLBA*) component, (rather than the *Steppe_EMBA* groups suggested earlier) (Figure 1).

Figure 1



A sketch of the peopling history of South Asia. Depicting the full complexity of available reconstructions is not attempted. Placing of population labels does not indicate precise geographic location or range of the population in question. Rather we aim to highlight the essentials of the recent advancements in the field. We divide the scenario into three time horizons: Panels (a) before 10 000 BCE (pre agriculture era); (b) 10 000 BCE to 3000 BCE (agriculture era) and (c) 3000 BCE to prehistoric era/modern era. (iron age).

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