Lactase persistence alleles reveal ancestry of southern African Khoe pastoralists

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In a new study a team of researchers lead from Uppsala University show how lactase persistence variants tell the story about the ancestry of the Khoe people in southern Africa. The team concludes that pastoralist practices were brought to southern Africa by a small group of migrants from eastern Africa. The study is published in *Current Biology* today.

"This is really an exciting time for African genetics. Up until now, routes of human migration in Africa were inferred mostly based on linguistics and archaeology, now we can use genetics to test these hypotheses." says Dr. Carina Schlebusch at Uppsala University.

Lactase persistence is the ability to digest milk into adulthood. There are a number of different genetic variants associated with lactase persistence and they are heterogeneously distributed in global populations. These variants were likely targets of selection when some populations converted from hunter-gatherer to herder and/or farming lifestyles. Specific lactase persistence variants are associated with particular geographic regions and populations; however, they have not been extensively studied in southern Africa before.

The indigenous Khoe people in southern Africa have historically been pastoralists and they have previously been shown to be closely related southern Africa's San hunter-gatherers.. The archeological record in southern Africa, for example from domesticated animals and material artifacts, is particularly clear on demonstrating Khoe herding practices and population continuity in the southwestern Cape from about 2,000

years ago and onwards. In a new study, Uppsala University researchers together with South African researchers show that lactase persistence variants were at medium frequencies in the Khoe people, but at very low frequencies or absent among San hunter-gatherers.

Using hundreds of thousands of markers, the team was able to show that some 13 percent of the genomes among Khoe pastoralists trace their ancestry to eastern African pastoralist groups. The team concludes that pastoralist practices were brought to southern Africa by a small group of migrants from eastern Africa which was assimilated by local indigenous hunter-gatherers, adopting the pastoralist lifestyle.

"The spread of pastoralism to southern Africa has been debated for many decades. We show that the spread was mediated by migration from eastern Africa. It is remarkable that a small group of migrants likely had a very strong impact on the way-of-life of the ancestors of the Khoe people." says Professor Mattias Jakobsson, Uppsala University.

"This study will help to write the history of the Khoe people and place their story into a global context of population diffusion" says Dr. Carina Schlebusch.

The team sequenced the lactase persistence regulatory region in 267 individuals from 13 southern African populations (including decedents of hunter-gatherers, herders and mixed farmers), providing the first comprehensive study of the lactase persistence regulatory region in a large group of southern Africans.

More information: Breton et al., Lactase Persistence Alleles Reveal Partial East African Ancestry of Southern African Khoe Pastoralists, *Current Biology* (2014), dx.doi.org/10.1016/j.cub.2014.02.041