

# **Are there changes in the DNA of indigenous southern African populations as a result of exposure to infectious diseases introduced by non-natives?**

**Katharine Owers**

When populations interact for the first time, at least one group is usually exposed to new infectious diseases. This was the case in southern Africa, where hunter-gatherer populations native to the area were exposed to immigrant groups with various origins and lifestyles. The first group came from eastern Africa around 2000 years ago and brought with it the practice of herding animals. The second immigration was of farmers from western and central Africa around 1200 years ago. The most recent migration, and potentially the most important for bringing new infectious diseases to the region, was of European colonists, who began arriving around 350 years ago. While there are not records of disease epidemics from the earlier encounters due to lack of written records, we know from European reports during colonization that in some epidemics as much as 90% of certain native populations succumbed to diseases such as smallpox. We also know, however, that many of the effects of these epidemics, even during colonial times, were not recorded, making it hard to determine the full extent of their impacts on the native populations.

In addition to written records, we can use genetic information to recreate and understand population histories. Events such as disease epidemics are a source of natural selection, and these selective events create certain patterns in the DNA they affect. I used those patterns to investigate selective pressure from introduced infectious diseases in indigenous southern African populations. I compared two populations with relatively well-known amounts of contact with the groups migrating into southern Africa. One population, the Ju/'hoansi, has been isolated throughout much of its history and has had little contact with outside groups, whereas the ≠Khomani population has had contact with migrants from all of the groups.

I used two complementary methods to search for differences in selection in immune system genes between these two populations. The hypothesis was that I would find more evidence of selection in the ≠Khomani, which had more exposure to the new diseases and had therefore likely been under more selective pressure. The first method looked at DNA regions that most strongly showed the patterns indicative of selection, and then within those regions that had likely been under selection, focused on immune system genes. The second method had a broader focus, examining the patterns indicating selection in immune system genes and comparing the results to those for the whole genome. The combination of these two methods was designed to give both an idea of immune system genes under strongest selection and a sense of the overall status of selection in the immune system versus the genome as a whole.

While the two methods give different information, both indicated that there had been selection on immune genes in the ≠Khomani, confirming my hypothesis that due to extensive contact with external groups, that population had adapted to infectious disease exposure. For the Ju/'hoansi, my first method, which examined areas of strongest selection, resulted in no such regions that contained immune genes, indicating that the most strongly selected genes in that population were not related to immune function. In addition, the second method, comparing the immune system to the whole genome, gave conflicting results based on the specific pattern of selection examined. One such pattern indicated no difference between immune genes and the rest of the genome, where the other indicated less selection in the immune system. The negative signal from one method and lack of a clear signal from the other again confirms my hypothesis that the Ju/'hoansi had low selective pressure due to introduced infectious disease due to its isolated location.