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Aboriginal Australian genomes reveal Indian ancestry

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Flow of genes suggests continent was not isolated after all.

This scenario is the result of a large genetic analysis outlined today in the *Proceedings of the National Academy of Sciences*[1]. It contradicts a commonly held view that Australia had no contact with the rest of the world between the arrival of the first humans around 45,000 years ago and the coming of Europeans in the 18th century.

"Australia is thought to represent one of the earliest migrations for humans after they left Africa, but it seemed pretty isolated after that," says Mark Stoneking from the Max Planck Institute for Evolutionary Anthropology in Leipzig, who led the new research.

Irina Pugach, a postdoctoral researcher from Stoneking's lab, discovered signs of the Indian migration by comparing genetic variation across the entire genomes of 344 individuals. These included Aboriginal Australians from the Northern Territories, highlanders from Papua New Guinea, several populations from Southeast Asian and India, and a handful of people from the US and China.

She confirmed an ancient association between the genomes of Australians, New Guineans, and the Mamanwa – a Negrito group from the Philippines. These populations diverged from each other around 36,000 years ago, suggesting that they all descended from an early southward migration out of Africa.

But Pugach also found evidence of more recent gene flow from India and northern Australia

This genetic mingling coincided with the arrival of microliths — small stone tools that formed the tips of weapons — and the first appearance of the dingo, which most closely resembles Indian dogs. All of these changes may be related to the same migration.

"There have been very few genetic studies of Australians," Stoneking says, "and not anything like the dense genome-wide study we carried out." A few smaller studies of mitochondrial DNA and the Y-chromosome had hinted at recent gene flow between India and Australia[2,3], but an earlier genome-wide study missed it by not including any Indian populations[4], and a project that sequenced a full Aboriginal genome dismissed signs of gene flow from India as a spurious result[5].

Sheila van Holst Pellekaan, a geneticist at the University of New South Wales welcomes the new study, but cautions that it is "definitely not representative of Australia". "We know that substantial diversity exists in Australia and that different entries had not been ruled out," she says. "Gene flow is likely to have occurred prior to European colonization, but not sufficient to disperse through pre-existing peoples."

She wants to know if people from other parts of the country also show signs of the same gene flow that Stoneking's team found. But a legacy of distrust about biological research among Aboriginal groups means that genetic studies are viewed suspiciously and samples are hard to come by. Stoneking acknowledges the problem. "We and a lot of other people would be interested in getting more genetic samples," he says, "but it's very difficult to do that."

3 Redd, A.J., et al. *Curr. Biol.* **12**, 673-677 (2002).

4 McEvoy, B.P., et al. *Am. J. Hum. Genet.* **87**, 297–305 (2011).

5 Rasmussen, M., et al. *Science* **334**, 94 (2011).

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