Evidence for mitochondrial DNA recombination in a human population of island Melanesia: correction

**E. Hagelberg^{1*}, N. Goldman², P. Liò², S. Whelan², W. Schiefenhövel³, J. B. Clegg⁴
and D. K. Bowden⁵**

¹*Department of Biochemistry, University of Otago, PO Box 56, Dunedin, New Zealand*

²Department of Zoology, University of Cambridge, Downing Street, Cambridge CB2 3EJ, UK

³Max-Planck-Institute for Behavioural Physiology, Von-der-Tann Strasse 3, D-82346 Andechs, Germany

⁴MRC Molecular Haematology Unit, Institute of Molecular Medicine, University of Oxford, John Radcliffe Hospital, Oxford OX3 9DS, UK

⁵*Department of Anatomy, Monash University, Clayton, Victoria 3168, Australia*

We recently presented evidence of mitochondrial DNA recombination in humans based on the observation of a rare mutation in several unrelated human lineages in Nguna, a small island in Vanuatu, island Melanesia. Since then, the mutation has been shown to be an artefact caused by misalignment of the DNA sequences. Our previous conclusion, that the presence of a rare mutation on different haplotypic backgrounds was a consequence of genetic recombination, is no longer tenable for these data.

Keywords: human evolution; mitochondrial DNA; recombination; Melanesia; Polynesia

1. INTRODUCTION

In a recent article (Hagelberg *et al.* 1999), we presented evidence of recombination in human mitochondrial DNA (mtDNA). We sequenced a portion of the hypervariable control region of mtDNA in 452 humans from different locations in the western Pacific. The sequences fell into three distinct mtDNA clusters or haplotypes, which reflect the settlement history of the Pacific. The first type (I) was characterized by a previously described Polynesian motif, and variants thereof, and was observed in island and coastal areas. It exhibited low variability consistent with the recent expansion of the Polynesians. The second and third types (II and III) were found in people of the highlands of New Guinea and in island Melanesia and probably derive from the Pleistocene settlement of the

western Pacific. We detected a very rare mutation in people of the three lineages living in the small Melanesian island of Nguna. The occurrence of a rare mutation on three distinct haplotypic backgrounds is a highly unlikely event. We concluded that paternal leakage of mtDNA and subsequent recombination were the most likely explanations for this phenomenon.

Since then, we discovered that our Nguna data were incorrect. The mtDNA sequences of the Nguna population sample were shifted by ten nucleotides in the positions before 16 099. The nucleotide substitutions up to that point were misread by ten bases relative to the reference sequence (Anderson *et al.* 1981). The previously described C to T substitution at position 16 076 corresponds to a T to C substitution at 16 086. The individuals thought to carry the rare mutation at 16 076 are in fact

Table 1. *Mitochondrial DNA control region substitutions in 41 inhabitants of the island of Nguna in Vanuatu, island Melanesia*

[illegible]

* Author for correspondence (erika.hagelberg@stonebow.otago.ac.nz).

identical to the reference sequence at this position. However, the classification of the sequences into three major lineages still holds true and is unaffected by the misreading of the Nguna sequences.

2. CORRECTED DATA

The corrected data are presented in table 1. Our previous conclusion, that the parallel occurrence of a rare mutation on different haplotypic backgrounds was highly improbable and more likely to be the consequence

of genetic recombination, is no longer tenable for these data.

REFERENCES

Anderson, S. (and 13 others) 1981 Sequence organisation of the human mitochondrial genome. *Nature* **290**, 457–465.
Hagelberg, E., Goldman, N., Liò, P., Whelan, S., Shiefenhövel, W., Clegg, J. B. & Bowden, D. K. 1999 Evidence for mitochondrial DNA recombination in a human population of island Melanesia. *Proc. R. Soc. Lond. B* **266**, 485–492.