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Natural radioactivity and human mtDNA mutations

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A 10-km stretch of coast in Kerala (India) contains the world's highest level of natural radioactivity in a populated area, offering a unique opportunity to identify "radiogenic" DNA mutations and to compare them with "evolutionary" mutations. We have sampled DNA from a total of 988 individuals from 247 native families (covering 791 mtDNA transmissions).

Two-thirds of the samples were taken from the radioactive coastal strip, and one-third from the 3-km distant, non-radioactive hinterland as a control population. We sequenced the control region of their mitochondrial DNA, and found both point mutations and homopolymeric length changes between mothers and their offspring. In each mutation case, we confirmed maternity with a probability of $>99.15\%$ by typing nine autosomal loci.

Our study reveals three main results:

(a) The families living in the radioactive area have seven times (SD$\pm$1.02) more new point mutations than the control families.
(b) Strikingly, the new mutations primarily affected nucleotide positions previously identified as hypervariable in evolutionary studies, raising the possibility that evolutionary point mutations in human mtDNA are largely the direct or indirect result of radioactivity.
(c) Of central importance to medical, forensic, and evolutionary geneticists is the finding that none of the point mutations attained fixation in any individual, resolving the perceived conflict between "evolutionary" and "pedigree" mtDNA mutation rates.

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