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Mitochondrial DNA homoplasmy

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Somatic point mutations in mitochondrial DNA (mtDNA) have been linked to aging and cancer. Each cell contains a large number of mtDNA molecules, so any mutation requires a process of clonal expansion in order to reach homoplasmy (close to 100%) and to exert a phenotypic influence. In the April 16 Proceedings of the National Academy of Sciences, Nekhaeva *et al.* document the frequency of mtDNA mutations in human tissues (*Proc Natl Acad Sci USA* 2002, **99:**5521-5526). They employed a cell-by-cell sequence-specific method to study buccal epithelial cells and cardiomycoytes from aged individuals (up to 109 years old!). They present evidence for intercellular clonal expansion of mtDNA mutations in cells from aged tissues; they found that the rate of spread to homoplasmy was very rapid. The spectra of mtDNA mutations were different in the two cell types and probably reflect a difference in the mechanisms of clonal expansion. These results support the possible link between mtDNA mutations and aging, but suggest that mtDNA mutations in tumor cells probably appear by the same process as in normal cells.

References

1. Mitochondrial DNA mutation associated with aging and degenerative disease.

2. Facile detection of mitochondrial DNA mutations in tumors and bodily fluids.

3. *Proceedings of the National Academy of Sciences*, [http://www.pnas.org]

4. Marked replicative advantage of human mtDNA carrying a point mutation that causes the MELAS encephalomyopathy.