

題名:A prevalent POLG CAG microsatellite length allele in humans and African great apes.

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上傳時間:2009-10-02T08:52:31Z

摘要:Abstract

The human nuclear gene for the catalytic subunit of mitochondrial DNA polymerase γ (POLG) contains within its coding region a CAG microsatellite encoding a polyglutamine repeat. Previous studies demonstrated an association between length variation at this repeat and male infertility, suggesting a mechanism whereby the prevalent (CAG)₁₀ allele, which occurs at a frequency of >80% in different populations, could be maintained by selection. Sequence analysis of the POLG CAG microsatellite region of more than 1000 human chromosomes reveals that virtually all allelic variation at the locus is accounted for by length variation of the CAG repeat. Analysis of POLG from African great apes shows that a prevalent length allele is present in each species, although its exact length is species-specific.

In common chimpanzee (*Pan troglodytes*) a number of different sequence variants contribute to the prevalent length allele, strongly supporting the idea that the length of the POLG microsatellite region, rather than its exact nucleotide or amino acid sequence, is what is maintained. Analysis of POLG in other primates indicates that the repeat has expanded from a shorter, glutamine-rich sequence, present in the common ancestor of Old and New World monkeys.