

**P018** Germline and somatic mutation at a human  
Y-specific minisatellite

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The human Y-specific minisatellite, MSY1, is haploid, and exempt from inter-allelic mutation processes, yet maintains a very high degree of structural diversity. Analysis of MSY1 alleles within the framework of the Y phylogeny suggests a role in mutation for gene conversion, operating either intrahelically following slipped strand mispairing, or interhelically via unequal sister chromatid exchange. Using a single-molecule PCR approach, mutation in matched sperm and blood samples has been studied. Sperm mutation rate was ~3%, and mutants fell into three classes: (i) small-scale length changes; (ii) internal structural changes, in which adjacent blocks of repeats respectively lose and gain the same number of repeats, while preserving overall allele length (isometric); and (iii) a single example of an isometric mutant with a more complex internal structural change. In blood DNA, the rate of mutation is similar (2.5%), but the spectrum is different, with 80% belonging to class (iii). The mutation spectrum in sperm DNA is consistent with processes inferred from the phylogeny, supporting a role for gene conversion, while sperm/blood differences suggest that mutational pathways in germline and soma are distinct.