Partial duplication at AZFc on the Y chromosome is a risk factor for impaired spermatogenesis in Han Chinese in Taiwan

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摘要

Abstract

The Azoospermia Factor c (AZFc) region on the Y chromosome long arm is one of the least stable regions in the human genome. It consists almost entirely of very long repeats and is prone to rearrangement. Numerous structures at AZFc have been identified, and some of them have been reported to be associated with male infertility. We screened 580 Han Chinese in Taiwan for AZFc deletion and duplication using three PCR assays, and characterized the DAZ genes in selected subjects with additional Southern analyses. About 9.5% of our subjects have AZFc partial deletion, 2.8% have partial deletion followed by duplication, and 1.7% have partial duplication. The overall rearrangement frequencies vary significantly between different Y chromosome haplogroups (Yhgs), ranging from 2.9% in O3e to 100% in N and Q. All individuals in Yhg-N lack the sY1191 marker, but one out of three of them actually have four DAZ genes, indicating further duplication after the b2/b3 deletion. Our additional screening of 142 oligospermic men and 107 fertile controls found no significant difference in the frequencies of the gr/gr and the b2/b3 deletion. However, the frequency of AZFc partial duplication in the infertile group (7.0%) was significantly higher than that in the fertile control group (0.9%) and the general Taiwanese population (1.7%). Our results indicate that AZFc partial deletion and partial duplication are common polymorphisms in Han Chinese, and that the AZFc partial duplication, but not the AZFc partial deletion, is a risk factor for male infertility in the Taiwanese population. 2007 Wiley-Liss, Inc.