



Homologous recombination between HERVs causes duplications in the AZFa region of men accidentally exposed to cesium-137 in Goiânia

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ABSTRACT. In September 1987, in Goiânia, Brazil, one of the most serious radiological accidents occurred at a radiation therapy unit involving a source of cesium-137. The current study examined the occurrence of possible germline mutations at the AZF region of the exposed men and in their male offspring. Genomic DNA samples of 16 individuals were analyzed for microdeletions. All exposed individuals amplified sequence tagged sites; however, sY84 and sY86 showed a duplication in 75% (12/16) of the exposed group. Exposed families designated as B and E showed a duplication of sY84 and sY86, both in the fathers and their sons. Fathers of families A, C, D, and F did not show a duplication in the AZF region, but their sons did. The children in A and D had duplications of sY84 and sY86, while children in families C and F had a duplication exclusively of sY84. Family G showed a duplication of sY84 in all three generations from grandfather to grandson. Two human endogenous retroviral sequences (HERV) exist in the AZFa region, and non-allelic recombination between these sequences could cause chromosomal rearrangements, such as deletions or duplications, and a mutational mechanism intrinsic to non-allelic recombination could be

increased by individual exposure to ionizing radiations from cesium-137. Consequently, the hotspots inside HERV mediated recombination in AZFa, and the duplication diversity was compatible with male fertility, since to date, none of the exposed individuals have demonstrated fertility disorders.

Key words: Y chromosome; Rearrangement; Ionizing radiation; Male fertility