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Thesis Abstract

Occurrence of mutations in loci linked to Y chromosome in the offspring born to individuals exposed to ionizing radiation

J.T. Arruda

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In September 1987, in Goiânia, GO, Brazil, one of the most serious radiological accidents occurred at a radiotherapy unit involving a source of cesium-137. An area of 2000 m^2 was contaminated and 249 people were exposed, both externally and internally, to substantial doses of ionizing radiation, resulting in four fatalities due to acute radiation syndrome. The current study examined the occurrence of possible mutations on the Y chromosome of the exposed men and their male offspring divided into two groups: A) eight accidentally exposed men and eight boys; B) twelve occupationally exposed men and sixteen boys; the control group was constituted by 8 men and 8 boys not exposed. DNA was isolated from peripheral blood lymphocytes and 30 loci (SRY, AMELY, ZFY, AZFa-Prox1, SY83, AZFa-Prox2, SY86, SY85, SY84, USP9Y, SY87, DBY, AZFa-Dist1, 12f2, AZFa-Dist2, UTYpe, SY106, SY124, SY127, SY134, SY135, SY143, SY1197, SY1291, SY1125, SY1054, YDAZ3, SY254, SY255, and RH65618) were amplified by the polymerase chain reaction. All DNA tests had a probability of paternity of at least 99.99%. All analyzed individuals amplified sequence tagged site; however, 4 fathers (8.4%) and 8 sons (21.2%) in the group A, and 3 fathers (7.1%) and 3 sons (63.3%) in the group B showed mutations. The total mutation rate was 0.11. The first generation of the accidentally exposed group showed 7 mutations in SY86, 12 mutations in SY84, and 1 mutation both in 12f2 and SY135. The first generation of the occupationally exposed group showed 2 mutations in SRY, AMELY, AZFa-Prox1, AZFa-Prox2, SY86, SY85, SY84, USP9Y, SY87, AZFa-Dist1, UTYpe, SY106, SY124, SY127, SY134, SY135, SY143, SY1125, SY1054, YDAZ3, SY254, SY255, and RH65618, and 4 mutations in 12f2. In the control group, only one son showed an SY84 deletion. Recombination events between repetitive regions are possibly the cause of the high incidence of *de novo* mutations on the Y chromosome. The mutations were possibly generated by intrinsic mechanisms that could have been increased by the ionizing radiation from cesium-137. The exposure to ionizing radiation from cesium-137 can be detected in offspring of exposed individuals, and the mutation rate can be attributed to radioactive exposure.

Key words: Recombination; Y chromosome; Ionizing radiations; Cesium-137

