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Effects of Robertsonian fusions on male reproduction: structural and ultrastructural analysis of the testis and epididymis

J. Repullés[1], M.A. Sans Fuentes[2,3], N. Medarde[4], J. Martínez[4], M. Ponsà[5], M.J. López Fuster[2], J. Ventura[4], A. Sanchez Chardi[1]

[1]Universitat Autònoma de Barcelona, Microscopy Facility, Cerdanyola del Vallès, Spain
[2]Universitat de Barcelona, Biologia animal, Barcelona, Spain
[3]University of Arizona, School of Geography and Development, Tucson, Spain
[4]Universitat Autònoma de Barcelona, Biologia animal, Cerdanyola del Vallès, Spain
[5]Universitat Autònoma de Barcelona, Biologia cel·lular, fisiologia i immunologia, Cerdanyola del Vallès, Spain

Alejandro.Sanchez.Chardi@uab.cat
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Robertsonian translocations (centric fusions between two telocentric chromosomes to form a metacentric one) can act as a mechanism of speciation reducing fertility, and have been described in some mammalian species, such as the house mouse, *Mus musculus domesticus*. To study the effect of these translocations on male reproduction, Robertsonian (Rb, 2n=28-31) and standard mice (St, 2n=40) from the Robertsonian polymorphism area of Barcelona (NE Spain) were analysed. Testis (tubular diameter, epithelium thickness, cell cycle arrest during spermatogenesis) and caput epididymis (percentage of abnormal spermatozoa) were evaluated using both light and electron microscopy.

The preliminary results comparing Rb and St mice can be summarized as follows: 1) the two groups did not show important differences in the structural morphology of the seminiferous tubules; 2) Rb mice showed an increase of cell cycle arrest (mainly ending in apoptosis; Figure 1) during spermatogenesis in comparison with St mice; 3) Rb mice had more mature spermatozoa with ultrastructural abnormalities in the head and the tail (Figure 2).

These results may be explained by the increase of defects during meiosis (e.g. impairment in homologous chromosome pairing) in Rb mice, producing an increase of unviable germ cells, and a subsequent decrease of fertility. These results point out the relationship between Rb reorganizations and both germ cell death and gamete anomalies. Our results using microscopy are concordant with previous studies on the Robertsonian zone of Barcelona, which indicate that in Rb mice the percentage of viable gametes is lower and the spermatogenesis less efficient than in St mice. All these findings suggest that in the study area Rb translocations reduce gene flow among populations as a result of a decrease in fertility in Rb mice with a relatively high number of metacentric chromosomes.

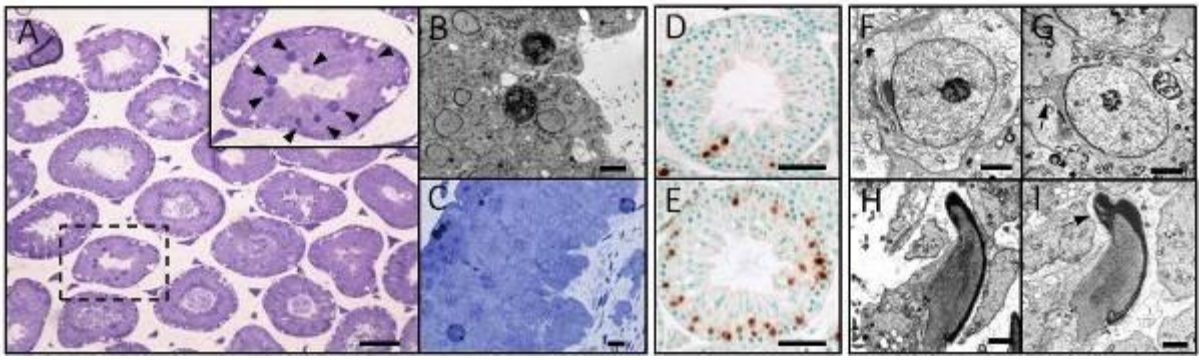

Figure 1. Representative images of testicles of Rb mice showing cell degeneration of tubules (A, B, C) mainly due to apoptosis (arrowheads). Apoptosis was confirmed by TUNEL assay (D: St, E: Rb). F-I micrographs showed normal round (F) and elongated (H) spermatid and abnormal round (G) and elongated (I) spermatids with acrosomal alterations (arrow). Bars: A=100µm; B=5µm; C=10µm; D, E=50µm; F-I = 2µm.

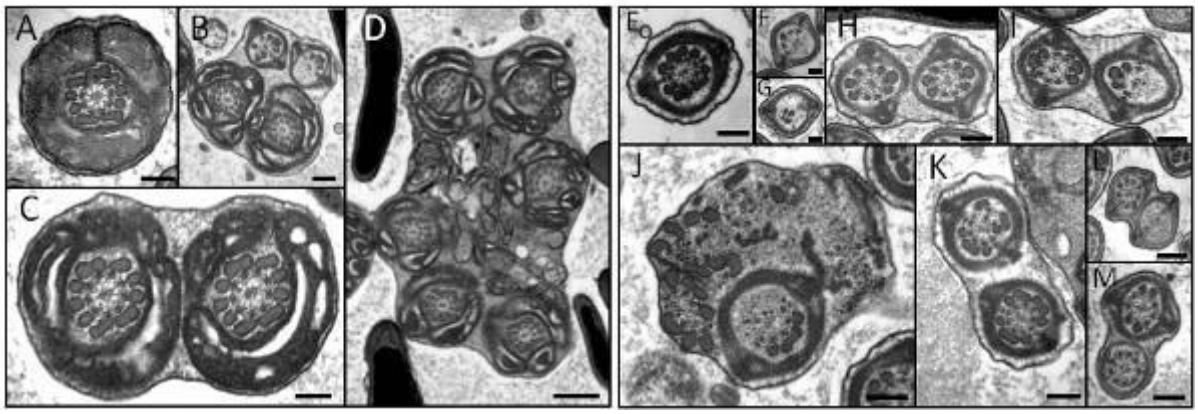

Figure 2. Representative micrographs showing the normal structure (A, E) and the most common ultrastructural alterations in tails of mice sperm. These abnormalities were significantly more frequent in Rb mice and were mainly based in two or more tail sections embedded in the same plasma membrane (C, D, H, M), abnormal microtubule number (F) or organization (G) and multiple alterations (B, I, J, K, L). Bars: A-C, E, H-M = 200nm; D=500nm; F, G = 100nm