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ASOCIACIÓN MADRILEÑA
PARA EL SÍNDROME DE
PRADER-WILLI



FERTILITY AND INFERTILITY IN PRADER-WILLI SYNDROME TESTICULAR HISTOLOGY IN NINE MALES WITH CRYPTORCHIDISM

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INTRODUCTION: Hypogonadism leading to genital hypoplasia and delayed or incomplete gonadal maturation is a central feature of the Prader-Willi syndrome (PWS). Cryptorchidism is found in 93% of the PWS patients and is considered to be a phenotypic criterion. Infertility is also thought to be a consistent characteristic in males with PWS. Nevertheless data on spermatogenesis are lacking. In normal males who undergo orchidopexy for cryptorchidism, there is a correlation between prepubertal lesions on testicular biopsies and the degree of spermatogenesis in postpubertal specimens (Nistal et al., 2000). To predict future spermatogenesis we analysed testicular histology in 8 prepubertal boys with cryptorchidism and a molecularly confirmed diagnosis of PWS. In addition we analysed the testicular histology in one adult male with cryptorchidism and torsio testis.

MATERIAL AND METHODS: We describe the testicular histology in 8 boys (6 with a deletion, 2 with uniparental maternal disomy) who underwent orchidopexy (13 testes) for cryptorchidism (age at time of biopsy: 16 months to 14 years) and in one adult male who underwent orchidectomy for cryptorchidism with torsio testis. On the basis of this testicular biopsy, prepubertal undescended testes were classified into four Nistal categories (Nistal et al., 1980) according to the mean tubular diameter (MTD), the tubular fertility index (TFI: the average percentage of tubules showing germ cells) and the Sertoli cell index (STI: number of Sertoli cells per cross-sectioned tubule): Nistal category I: normal MTD, TFI >50%, SCI normal; Nistal category II: slight to marked reduced MTD, TFI 30-50%, SCI normal; Nistal category III: MTD severe and diffuse reduced, TFI <30%, SCI severely reduced; Nistal category IV: normal MTD, variable TFI, diffuse Sertoli cell hyperplasia.

RESULTS: Out of the eight prepubertal boys, two (25%) showed a Nistal score of I, one (12.5%) showed a Nistal score of II and five males (62.5%) showed a Nistal score of III. The testis of the adult male showed a Sertoli cell nodule, vacuolised Leydig cell, peritubular hyalinisation and small tubuli.

CONCLUSION: PWS appears to be a heterogeneous disorder with respect to testicular histology. Although a great part (62.5%) of the prepubertal boys with cryptorchidism show absence of spermatogonia, 25% of the boys have a normal testicular histology. Future studies are necessary to evaluate the evolution of germ cells in males with the Prader-Willi syndrome and to correlate these data with testicular histology and spermiogram in adulthood.

References

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