Genetic Causes of Azoospermia Secretory in Our Midst

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Objective. Description of frequently genetic alterations found in infertile men with azoospermia secretory at our hospital and comparison with findings by other studies. Specifically, a significantly lower prevalence of microdeletions at Y Chromosome was observed. METHODS: This is a retrospective study aimed to evaluate the genetic behavior of men diagnosed as azoospermic during a two-year period in order to compare the results with frequencies reported worldwide. PATIENTS: 108 men were diagnosed with azoospermia secretory at Hospital Universitario Río Hortega de Valladolid during a two-year periods. Age ranged from 23 to 48 years old. RESULTS: 12% of men from the West Area of Valladolid who underwent a test for infertility during suffered azoospermia secretory, a percentage similar to that found in other populations and studies. Incidence of microdeletions at Y Chromosome (1.85%) was significantly lower than what has been reported by other authors (8%-12%). Only two azoospermic men out of 108 had microdeletions at Y Chromosome. Chromosomal abnormalities (whether numerical or structural) were present in 6.48% of those diagnosed as azoospermic (7 patients out of 108). Incidence of heterozygous mutations at CFTR Gene was 5.55% (6 patients out of 108), three of them associated with the 5T variant. The most frequent genotype 7T/7T accounted for 76% of the sample, and no cases with 5T/5T genotype were detected. If an 8% worldwide prevalence of microdeletions at Y Chromosome is considered, then p value = 0.0053. If a 12% worldwide prevalence of microdeletions at Y Chromosome is considered, then p value = 0.0001. CONCLUSIONS: Incidence of microdeletions at Y Chromosome differs across studies and authors. Genetic studies are variable and depend much on the patients' race. It must be taken into account that all but two of the patients in our study population were white-caucasian.