CYTOGENETIC STUDY IN MEN FROM THE POPULATION OF THE REPUBLIC OF MOLDOVA

Stela RACOVITA[1], Veaceslav MOSIN[1], Mariana SPRINCEAN[1,2]

[1]*Nicolae Testemitanu* State University of Medicine and Pharmacy, Chisinau, Republic of Moldova
[2]Institute of Mother and Child, Chisinau, Republic of Moldova

Autor corespondent: Stela Racovita, e-mail: stela.racovita@usmf.md

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Introduction. Globally, it is estimated that 15% of couples experience infertility, with male factors contributing to half of these cases. The causes of male infertility are highly diverse, often stemming from disorders in spermatogenesis, which are clinically characterized by severe oligozoospermia and azoospermia. Genetic factors account for approximately 30% of male infertility cases associated with spermatogenesis disorders. Among the numerous genetic causes of spermatogenic insufficiency, chromosomal anomalies are particularly significant from a clinical standpoint. Approximately 15% of men with non-obstructive azoospermia and 4% of men with severe oligozoospermia have chromosomal abnormalities.

The aim of this study is to evaluate the profile of chromosomal variations in male infertility with severe oligozoospermia and azoospermia, in order to confirm the importance of exploring their cytogenetics for both diagnosis and treatment, as well as for assessment and prognosis.

Material and methods. The study involved a retrospective descriptive analysis of cytogenetic results in men with oligozoospermia and azoospermia in the Moldovan population from 2014 to 2022. The participants were individuals from infertile couples who sought assistance at the National Center for Reproductive Health and Medical Genetics. The patient group comprised 32 individuals with oligozoospermia and 156 with azoospermia. All patients underwent cytogenetic analysis using the classical G-banding technique on peripheral blood lymphocytes. Results were reported using the nomenclature outlined in the 2016 International System of Cytogenetic Nomenclature (ISCN).

Results. The average age of men was 33.8±5.3 years, (95% CI: 32.7-34.9; median 33.0) (percentile 25 – 75: 30.0-36.0). Among the 156 men with azoospermia, 101 (64.7%) had a normal karyotype of 46,XY, while variations in the number or structure of chromosomes were observed in 55 individuals (35.3%; 95% CI: 27.8-42.8). Of these, 43 patients (27.6%) exhibited variations in the X or Y sex chromosomes, and 12 patients (7.7%) showed variations in the autosomal chromosomes. Among the 43 azoospermic patients with sex chromosomal abnormalities, 32 cases involved chromosomal number abnormalities, and 11 cases involved structural variations. Among the number of chromosomal abnormalities, 29 cases were attributed to X aneuploidy (Klinefelter Syndrome), 2 cases were mosaic 45,X/46,XY, and one case presented a 46,XX karyotype in a male. The structural variations of the sex chromosomes included 9 cases of duplications of the distal arm of the Y chromosome (Yqh+) and 3 cases of deletions of the distal arm of the same chromosome (Yqh-). Among the 32 men with oligozoospermia, 28 (87.5%; 95%CI: 76.0-99.0) had a normal karyotype of 46,XY, while 4 (12.5%; 95% CI: 1.0-24.0) exhibited variations in the sex chromosomes.

Conclusions. In this study, the prevalence of chromosomal abnormalities identified in azoospermia was 35.3% and in oligozoospermia was 12.5%. Given the high frequency of chromosomal abnormalities in men with infertility, as well as the genetic risks for future generations, it is important to assess the profile of chromosomal variations before resorting to Assisted Reproduction Techniques, for diagnosis, treatment, as well as assessment and prognosis.