CYTOGENETIC ANALYSIS IN MALES WITH AZOOSPERMIA

Stela Racoviță [1], Moșin Veaceslav [1], Capcelea Svetlana [1], Mișina Ana [2], Cemortan Igor [1], Sprincean Mariana[1,2]

[1]State University of Medicine and Pharmacy, *Nicolae Testemițanu*

[2]Institute of Mother and Child, Chișinau, Republic of Moldova

BACKGROUND

➢Male infertility is a heterogeneous disorder with a substantial genetic basis.

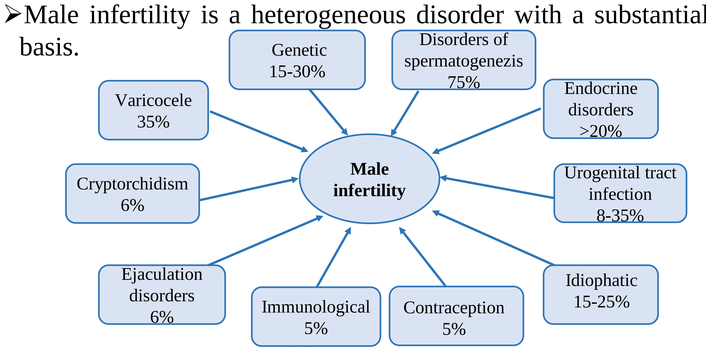

Figure1: Distribution of the most common causes of male infertility

➢The most common genetic causes of male infertility are chromosomal anomalies and microdeletions of the azoospermia factor (AZF).

➢The frequency of these chromosomal anomalies increases in azoospermic men.

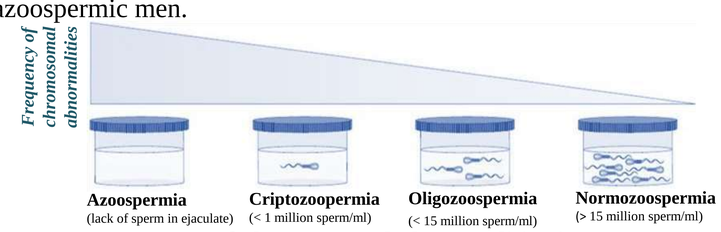

Figure 2: Frequency of chromosomal abnormalities in spermatogenesis disorders

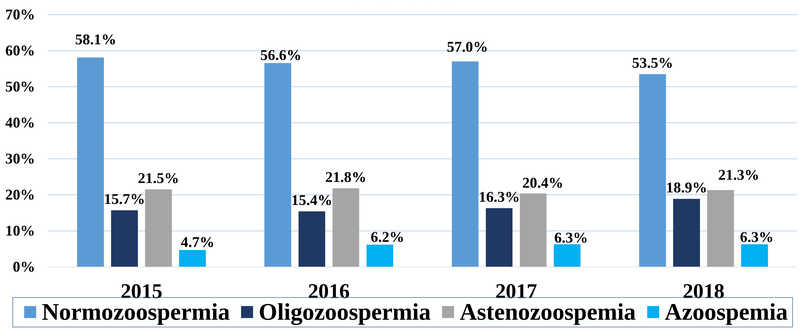

Figure 3: Semen structure to infertile men during 2015-2018 from RM

THE PURPOSE

➢ To assess chromosomal variations in males with azoospermia in order to confirm the importance of the cytogenetic testing for diagnosis and treatment assessment.

METHODS

➢ We performed cytogenetic analysis in a group of 128 infertile men with azoospermia from the Republic of Moldova during 2013-2018 period.

➢ Karyotyping was performed on peripheral blood lymphocytes according to standard methods G-banding of metaphase chromosomes. For reporting the results, the 2016 International System of Cytogenetic Nomenclature was used.

RESULTS

➢ 128 men with azoospermia were cytogenetically investigated in 2013-2018 at the department of the National Center for Reproductive Health and Medical Genetics (Table 1).

Table 1. Distribution of chromosomal abnormalities in men with azoospermia, years 2013-2018

Years	Men with azoospemia Abs. No.	The average age/years	46,XY	Karyotype with chromosomal variations
2013	22	35	15	6
2014	23	35	13	8
2015	22	33	12	10
2016	21	33	12	9
2017	22	26	13	8
2018	18	32	13	5
Total	128	32	80	48

➢ We identified that from 128 azoospermic cases, 80 (62%) had normal karyotype (46,XY) and 48 (38%) showed variations in the number or structure of chromosomes. 38 patients (30%) showed variations in the X or Y sex chromosomes, and 10 patients (8%) had variations in the autosomal chromosomes (Figure 4, Table 1, 2, 3).

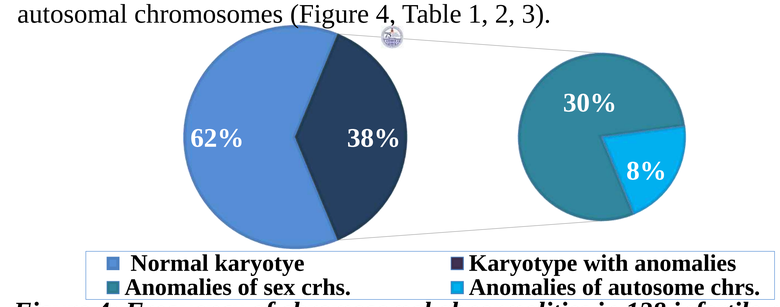

Figure 4: Frequency of chromosomal abnormalities in 128 infertile men

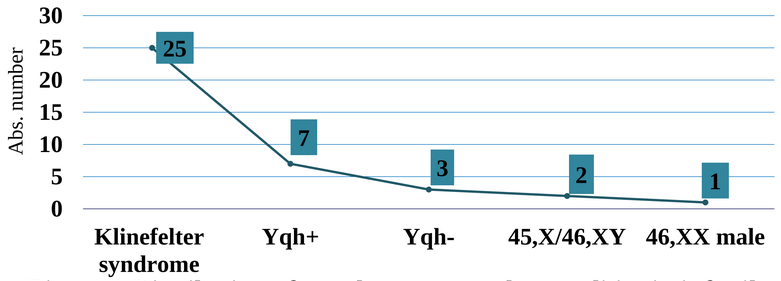

Figure 5:Distribution of sex chromosome abnormalities in infertile men

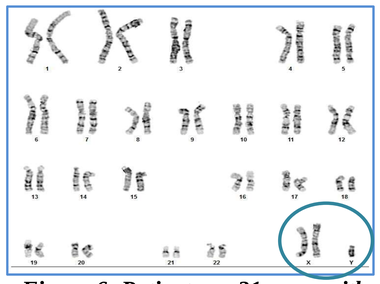

Figure 6: Patient age 31 years with Klinefelter Syndrome 47,XXY

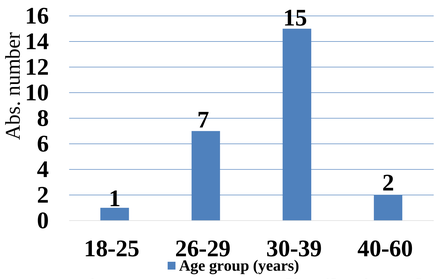

Figure 7: Age group distribution of Klinefelter Syndrome in infertile men

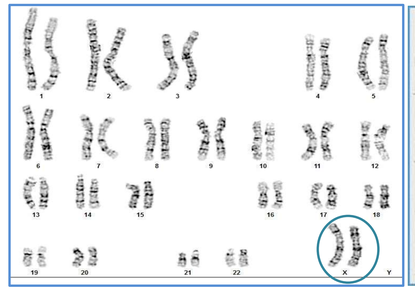

Figure 8: Patient age 30 years with 46,XX karyotype in male

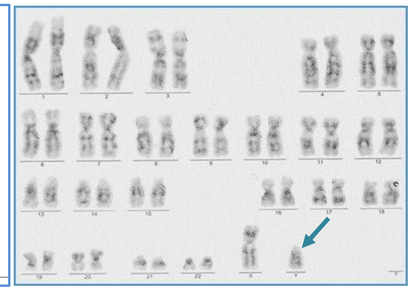

Figure 9: Patient age 29 years with 46,XYqh+ karyotype

Table 2: Specification of autosome chromozome abnormalities in 128 infertile men

Karyotype	Abs. No. (n=10)
Reciprocal translocations	**5**
46,XY,der(7),t(12;7)(12qter::7p21→pter)	1
46,XY,der(15), t(13;15) (13qter ::15q23 →qter)	1
46,XY,t(8;7)(8qter ::7q336 →qter)	1
46,XY-15-12,+der(15),+rec(12;15),t(13;12)7p+	1
46,XY,der(5),t(9;5)(9pter::5q23.3→qter)	1
Chromosomial polymorphism	**3**
46,XY,15 ps+	1
46,XY,14 ps+	1
46,XY,13 ps+	1
Inversions	**1**
46,XYinv(9)(p11q12)	1
Partial duplications	**1**
46,XY,1q+	1

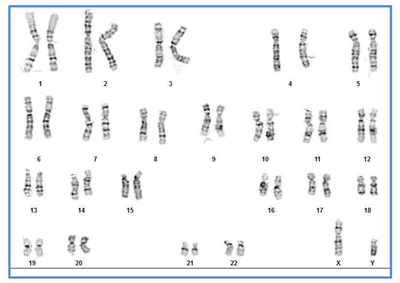

Figure 10. Pathient age 44 years with karyotype 46,XY,15ps+

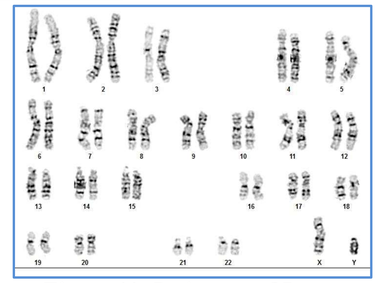

Figure 11: Patient age 35 years diagnosed with 46,XY, inv(9)(p11q12)

CONCLUSIONS

➢ Cytogenetic investigations for men with severely affected spermiogram is important for the etiologic diagnosis of male infertility with clinical relevant in treatment, as well as assessment and prognosis. The occurrence (38%) of chromosomal variations among infertile males strongly suggests genetic testing prior to ART.

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