EVALUATION OF MALE INFERTILITY MOLECULAR ASPECTS IN LATVIAN POPULATION

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Background

Some aspects of male infertility could be explained analysing Y-chromosomal and autosomal genes that are involved in spermatogenesis process.

The aim

To study molecular-genetic aspects of idiopathic male infertility in Latvian population.

Materials and methods

A study encompassed 100 idiopathic infertile men and 153 individuals of control group.

Y chromosome microdeletions were analysed by two multiplex PCR. Y chromosome haplogroups (Y-Hg) were detected using appropriate biallelic markers. CFTR gene mutations delF508, R117H and IVS8 polymorphisms (poly-T and poly-TG) analysis was performed by PCR and followed by RFLP and sequencing.

Results

Y-chromosome microdeletions were detected in 5% (5 cases of 100: 3 cases AZFc; 2 cases AZFa+b+c) (Figure 1).

Y-Hg analysis showed that Hg N3a1 and Hg R1a1 were less frequent in infertile male group compared to control group, however Hg K* was predominantly found in infertile male ($p<0.001$; *Bonferroni* corection) (Table 1).

Analysis of *CFTR* gene mutations delF508 and R117H and IVS8 polymorphisms (Figure 2) could not confirm association with infertility.

Figure 1. Data of Y chromosome microdeletion analysis in patients with AZF region microdeletions

Black boxes, STS marker absent – microdeletion of corresponding AZF region/ locus (Multiplex A and B PCR results). PAR1 and PAR2 - pseudoautosomal regions.

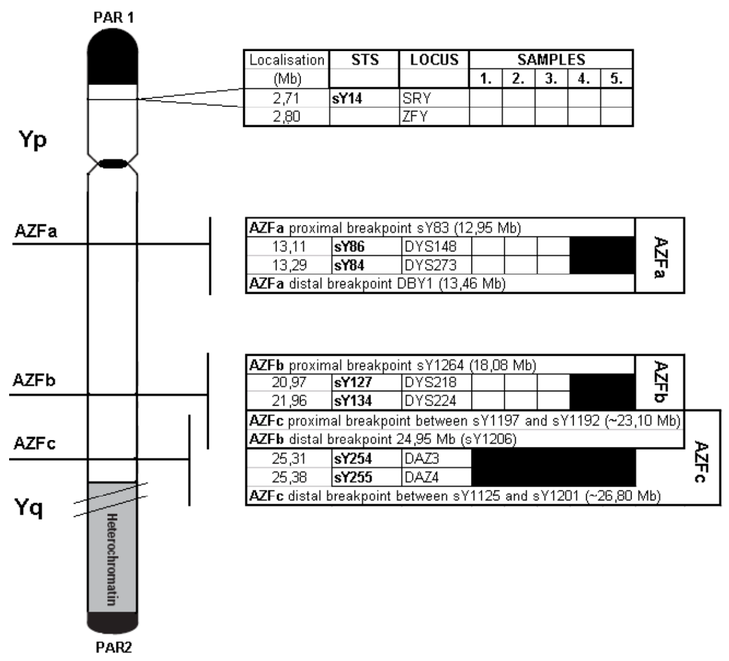
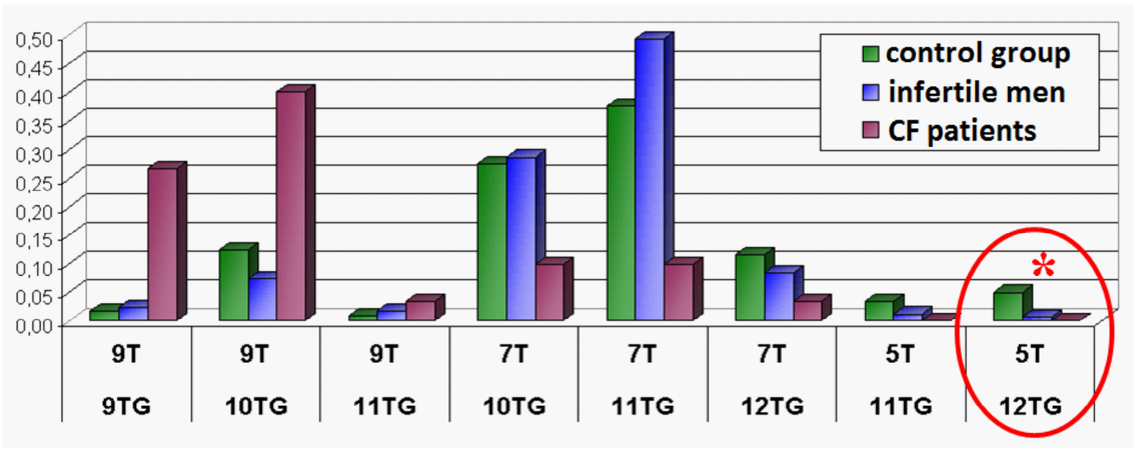


Table 1. Incidence of Y chromosome haplogroups among infertile males and control group

Haplogroups (Hg)	Incidence of haplogroups (%)		p
	Infertile males n = 79	Control group n = 153	
N3a1	**24 (30.4%)**	**65 (42.5%)**	**0.223**
R1a1	**12 (15.2%)**	**60 (39.2%)**	**0.005**
I	**2 (2.5%)**	**13 (8.5%)**	**0.097**
K* (without N3 and R1a)	**20 (25.3%)**	**10 (6.5%)**	**< 0.001**
Other (M9 C; DE* and F*:G, J)	**21 (26.6%)**	**5 (3.3%)**	**< 0.001**

Figure 2. Polymorphisms of CFTR gene poly-T and poly-TG variants in the analysed samples



* statistically significant differences (95% CI=0.005-0.099; χ^2=3.635, p=0.022)

Conclusions

1. Frequency of Y chromosome microdeletions in males with idiopathic infertility is 5%.
2. Y chromosome Hg K* may be associated with male infertility.
3. *CFTR* gene mutations delF508, R117H, and IVS8 polymorphisms do not affect process of spermatogenesis directly.

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