AZF midrodeletions screening in infertile men of the Portuguese population

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Analysis of genetic conditions associated with male infertility is, at present days, restricted to chromosome analysis, AZF Y-chromosome microdeletions screening, and to patients with hypogonadotrophic hypogonadism or with congenital absence of the vas deferens. Among different populations AZF microdeletions can explain 10-15% of the infertile phenotype of azoospermic men and 2-5% of oligozoospermic men.

Here we present the results of AZF deletions screening performed in a selected group of infertile Portuguese men with idiopathic non-obstructive azoospermia or with oligozoospermia (men with other causes of male infertility, endocrinological alterations, varicocele, criptorquidism, professional risk factors, autosomal chromosomal abnormalities, were excluded for this study).

Analysis was performed by Multiplex-PCR using specific STS for the three AZF regions. Microdeletion breakpoints were confirmed using a second multiplex-PCR. We analysed 865 infertile men (270 azospermic and 595 oligozoospermic with [spermatozoa]<$5x10^6$/mL) and 300 DNA samples obtained from fertile men of the Portuguese population.

While AZF microdeletions were found in 27 azoospermic (**10.0%**) and in 22 oligozoospermic men (**3.7%**), in fertile men no microdeletions were detected.

Patient	AZF region deleted (%)					
	AZFc	**AZFb**	**AZFa**	**AZFc+b**	**AZFc+b+a**	**Total**
Azoospermic	**9(3.3%)**	**4(1.5%)**	**1(0,4%)**	**10(3.7%)**	**3(1.1%)**	**27(10.0%)**
Oligozoospermic	**22(3.7%)**	**0**	**0**	**0**	**0**	**22(3.7%)**

Our results demonstrate that AZF microdeletions are frequent among males with the infertile phenotype described above. The regions absent have prognostic value for the clinical decision and patients treatment. Genetic counselling is recommended to all patients with AZFdel. AZF**c**del will be obligatorily transmitted to all male offspring by ICSI, which will seriously impair their fertility.