Chromosomal disorders and male infertility

Simão L.[1], Brito F[1], Caetano I[2], Marques B[1], Pedro S[1], Alves C[1], Silva M[1], Serafim S[1], Ambrósio P[1], Geraldes MC[1], Gonçalves J[2], Correia H[1].

[1]Unidade de Citogenética, Departamento de Genética Humana, Instituto Nacional de Saúde Dr. Ricardo Jorge, INSA,IP, Lisboa. [2]Unidade de Genética Molecular, Departamento de Genética Humana, Instituto Nacional de Saúde Dr. Ricardo Jorge, INSA,IP, Lisboa.

Male factor infertility is considered a complex disorder with a largely unknown etiology that affects about 7% of men. In general, genetic abnormalities account for 15%-30% of condition and Y chromosome microdeletions are also frequent.
The study, based on our casuistic, aimed at contributing to a better understanding of the genetic causes of infertility.

A group of 410 idiopathic infertile men with non-obstructive azoospermia, oligozoospermia, or unknown semen quality (based on clinical evaluation and/or sperm counts) was retrospectively selected. Conventional karyotype was performed in all samples; Y microdeletion screen was performed in 247 samples.

Forty two abnormal karyotypes (10.2%) were found, indicating an elevated frequency of chromosome abnormalities among the selected infertile men, as compared to that of newborn populations (≈0.4%). This frequency is higher than that reported in most similar studies that pointed to frequencies ranging from 2.2%-14.3%.

Klinefelter´s syndrome was the most common chromosome disorder (4.9%). There were 18 cases with 47,XXY karyotype and 2 cases of mosaicism involving lines 47,XXY and 46,XY. Reciprocal translocations were identified in 10 cases (2.4%), particularly in men with unknown semen quality. Overall, reciprocal translocations have been found in approximately 1% of the infertile men and more commonly in azoospermics than in oligozoospermics. However, this type of association was not found in the present study.
On the other hand, Y microdeletions were identified in 16/247 cases (6.5%), more frequently in azoospermics (13.3%, corresponding to 8/60 azoospermics). Among these 8 cases, 7 presented deletions at the AZFc region.
The marked presence of chromosomal abnormalities and Y microdeletions enphasizes the relevance of studying both factors in infertile men to improve genetic counseling, to allow the development of appropriate therapies, and to expand the knowledge about the ethiology of male infertility.

Table I – Number and frequency of chromosome abnormalities observed in 410 infertile men with azoospermia (AZO), oligozoospermia (OLIGO), oligo-asteno-teratozoospermia (OTA), cryptozoospermia (CRY) and infertile men with no indicated semen quality (InferNIQ).

Chromosomal abnormalities	AZO	OLIGO	OTA	CRY	OLIGO +OTA +CRY	InferNIQ	TOTAL 42
Sex chromosomes	15		3	1	4	8	**27**
47,XXY	9		1	1	2	7	18
mos 47,XXY[41]/46,XY[9]	1						1
mos 47,XXY[2]/46,XY[48]			1		1		1
47,XYY			1		1		1
mos 47,XXY[13]/46,XX[37]	1						1
46,X,der(X)t(X;Y)(p22.31;p11.2).ish der(X)t(X;Y)(p22.31;p11.2)(SRY+)	1						1
46,X,der(X)t(X;Y)(p22.33;p11.3).ish der (X)t(X;Y)(p22.33;p11.3)(wcpY+;SRY+)	1						1
46,X,+mar.ish der(Y)(wcpY+,DYZ3+)						1	1
46,X,+mar, inv(5)(p14.2p15.2)[32]/45,X,inv(5)(p14.2p15.2)[18].ish i(Y)(p10)(SRY++,DYZ3+, inv(5)(wcp5+)	1						1
mos 45,X[34]/46,X,idic(Y)(q11.221)[16]	1						1
Autosomes	1	2	1	0	3	11	**15**
46,XY,t(1;3)(q42.3;q26.2)						1	1
46,XY,t(1;10)(p22.1;q25.2)						1	1
46,XY,t(4;8)(q27;q11.23)		1			1		1
46,XY,t(4;22)(p10;q10)						1	1
46,XY,t(7;22)(q11.23;q11.2)						1	1
46,XY,t(8;14)(p23.3;q11.2)						1	1
45,XY,rob(13;14)(q10;q10)						2	2
46,XY,rob(13;14)+mar.ish der(16)(wcp16+)		1			1		1
46,XY,t(4;14;15)(q22;q21;q21.39).ish der(4)t(14;15)(wcp15+),der(14)t(4;14)(wcp4+), der(15)t(14;15)(wcp14+)			1		1		1
Total tranlocations	0	2	1	0	3	7	**10**
46,XY,inv(2)(p11.2q13)						1	1
46,XY,inv(8)(q21.2q22.3)						1	1
46,XY,inv(14)(q13q22).ish inv(14)(p21.1)(RP11-388M7+)(q21.3)(RP11-,168D12+)						1	1
46,XY,dup(8)(p23.1p23.1).ish (wcp8+)	1						1
47,XY,+mar.ish der(14/22)(D14Z1/D22Z1+)						1	1
Other structural	1	0	0	0	0	4	**5**
Total							42
Y deletions*							**30**
AZFa	4						4
AZFb	3					1	3
AZFc	6		1	1		4	12
gr/gr AZFc			1				1
SRY absence (and in X chromosome)	2						2

**some are presents in the same case*

Chromosomal abnormalities	Case n.	Foto	Indicação	FISH	Microd Y
Sex chromosomes					
47,XXY 80750 349769,307991, 406016, 175993,164370					
mos 47,XXY[41]/46,XY[9]	164370	-	azoosp	-	
mos 47,XXY[2]/46,XY[48]	146671	-	OTA	-	
Total mos XXY/XY					
47,XYY	116498	-	OTA	-	
mos 47,XXY[13]/46,XX[37]	136917	tem	azoosp	-	
46,X,der(X)t(X;Y)(p22.31;p11.2).ish der(X)t(X;Y)(p22.31;p11.2)(SRY+)	137431	Tem	Azoosp	Sonda p/ SRY Sinal no X	del a,b,c SRY no X
46,X,der(X)t(X;Y)(p22.33;p11.3).ish der (X)t(X;Y)(p22.33;p11.3)(wcpY+;SRY+)	79446				
46,X,+mar.ish der(Y)(wcpY+,DYZ3+)	118779	Não	Infert	wcpY e DYZ3	
46,X,+mar, inv(5)(p14.2p15.2)[32]/45,X,inv(5)(p14.2p15.2)[18].ish i(Y)(p10)(SRY++,DYZ3+, inv(5)(wcp5+)	133654	Tem		Wcp 5, DYZ3, SRY	del a,b,c
46,X,inv(5)(p14.2p15.2),+mar[32]/45,X,inv(5)(p14.2p15.2)[18].ish i(Y)(p10)(SRY++,DYZ3+, inv(5)(wcp5+)					
mos 45,X[34]/46,X,idic(Y)(q11.221)[16]	368774				
Autosomes					
46,XY,t(1;3)(q42.3;q26.2)	343092				
46,XY,t(1;10)(p22.1;q25.2)	386857				
46,XY,t(4;8)(q27;q11.23)	182396	Tem	Oligo	Não	
46,XY,t(4;22)(p10;q10)	405321				
46,XY,t(7;22)(q11.23;q11.2)	156217	tem		Não	
46,XY,t(8;14)(p23.3;q11.2)	383373	Tem		Não	
45,XY,rob(13;14)(q10;q10)	65148, 399891				
46,XY,rob(13;14)+mar.ish der(16)(wcp16+)	125376	Não	Oligozo	Painel de sondasCent wcp16+	
46,XY,t(4;14;15)(q22;q21;q21.39).ish der(4)t(14;15)(wcp15+),der(14)t(4;14)(wcp4+), der(15)t(14;15)(wcp14+)	118775	Tem	OTA	Wcp 4; wcp 14, wcp 15	
Total tranlocations					
46,XY,inv(2)(p11.2q13)	271413				
46,XY,inv(8)(q21.2q22.3)	380104			Tem Array	
46,XY,inv(14)(q13q22).ish inv(14)(p21.1)(RP11-388M7+)(q21.3)(RP11-,168D12+)	203679	Não	infert	Várias sondas 14	
46,XY,dup(8)(p23.1p23.1).ish (wcp8+)	138967	Não	azoosp	Wcp8	
47,XY,+mar.ish der(14/22)(D14Z1/D22Z1+)	340571	Não		Sond reg cent 13/21 14/22, 5, X, Y, 2 e 12	
Other structural					
Total					
Y deletions*					
AZFa					
AZFb					
AZFc					
gr/gr AZFc					
SRY absence or presence in X chromosome					

**some are presents in the same case*

Bárbara imagens FISH c/ legendas

Chromosomal abnormalities	Case n.	Fotos	Indicação	FISH	Microd Y
46,X,der(X)t(X;Y)(p22.31;p11.2)	137431	Tem	Azoospermia	Sonda p/ SRY Sinal no X	del a,b,c SRY no X
	ou				
der(X)t(X;Y(p22.33;p11.3).ish der (X)t(X;Y)(p22.33;p11.3)(wcpY+,SRY+)	79446		Azoospermia	wcpY, SRY	del a,b,c SRY no X
46,X,+mar.ish der(Y)(wcpY+,DYZ3+)	118779	Não	Infertilidade	wcpY e DYZ3	
mar,inv46,X,+ (5)/45,X,inv(5). Ish i(Y)(p10)	133654	Tem	Azoospermia	Wcp 5, DYZ3, SRY	del a,b,c
46,XY,rob(13;14)+mar.ish der(16)(wcp16+)	125376	Não sei	Oligozoospermia	Painel de sondasCent wcp16+	
46,XY,inv(8)	380104			Tem array	
46,XY,t(4;14;15)(q22;q21;q21.39).ish der(4)t(14;15)(wcp15+),der(14)t(4;14)(wcp4+), der(15)(wcp14+)	118775	Tem	OTA	Wcp 4; wcp 14, wcp 15	
46,XY,inv(14)(q13q22)	203679	Não	infertilidade	Várias sondas 14	
46,XY,dup(8)(p23.1p23.1).ish wcp8+)	138967	Não	azoospermia	Wcp8	
47,XY,+mar.ish der(14/22)(D14Z1/D22Z1+)	340571	Não	Infertilidade	Sond reg cent 13/21 14/22, 5, X, Y, 2 e 12	

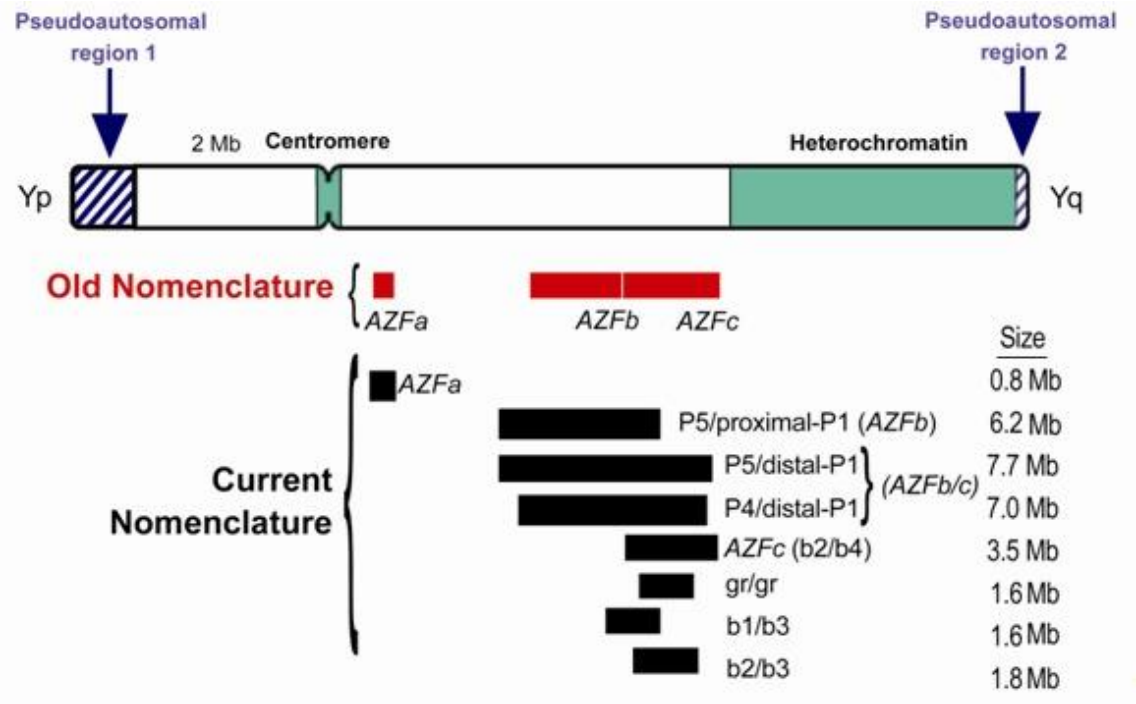


Figure – **Esteves, S. International Braz J Urol., 37(1):7-19**

Adaptado de Repping, S. et al., 2002, *Am J Hum Genet.* 2002; 71(4):906-922.

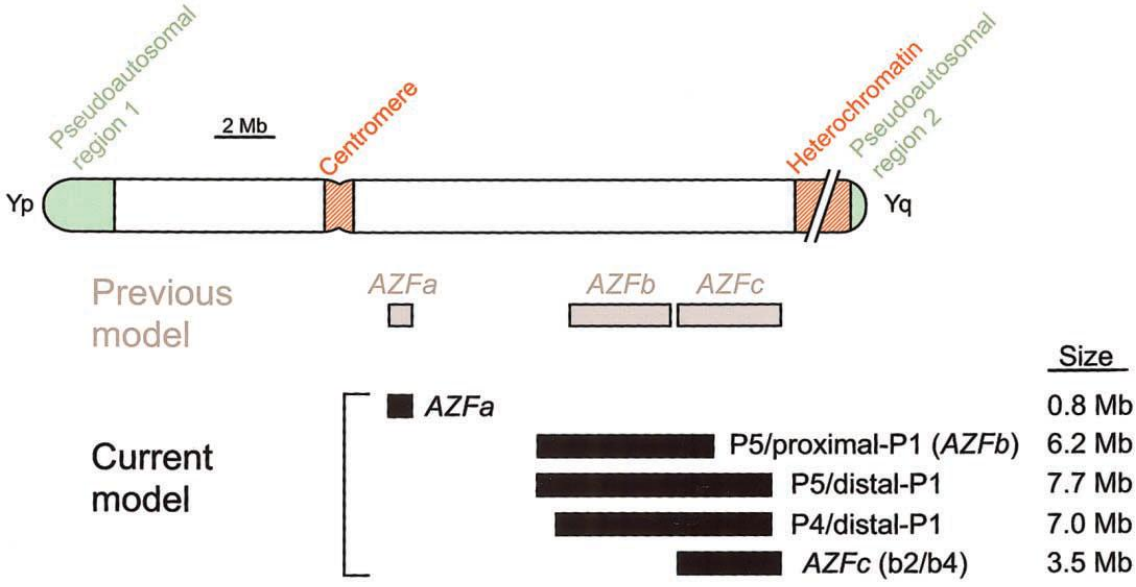


Figure 7 Previous model of recurrent, interstitial Y-chromosome deletions that cause infertility in men (Vogt et al. 1996) contrasted with current model.

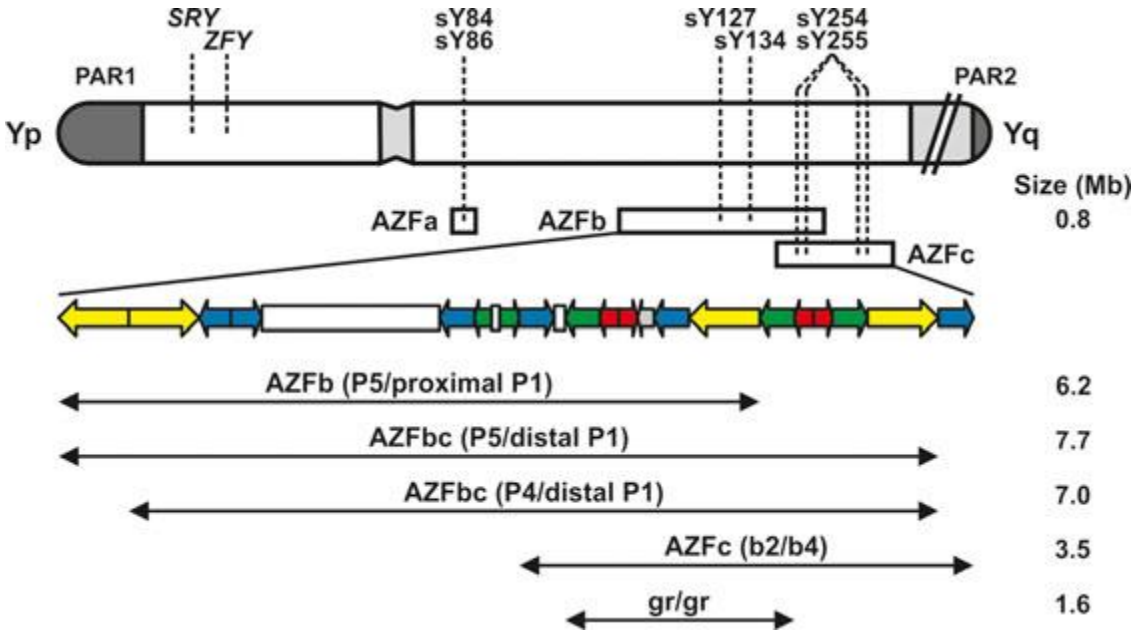


Figure - Schematic representation of the Y chromosome and the current microdeletion model (Repping et al., 2002). Repetitive sequences (colour coded palindromes) explain the origin of deletions in the AZFbc region by homologous recombination between identical sequences. The location of the STS primers suggested by the present guidelines is indicated by dashed lines. As four copies of the DAZ gene are normally present on the Y chromosome, the STS primers sY254, sY255 amplify four loci in AZFc. The AZFc (b2/b4) deletion is by far the most frequent type (~80%) of Y-chromosomal microdeletions found in men with severe oligo/azoospermia

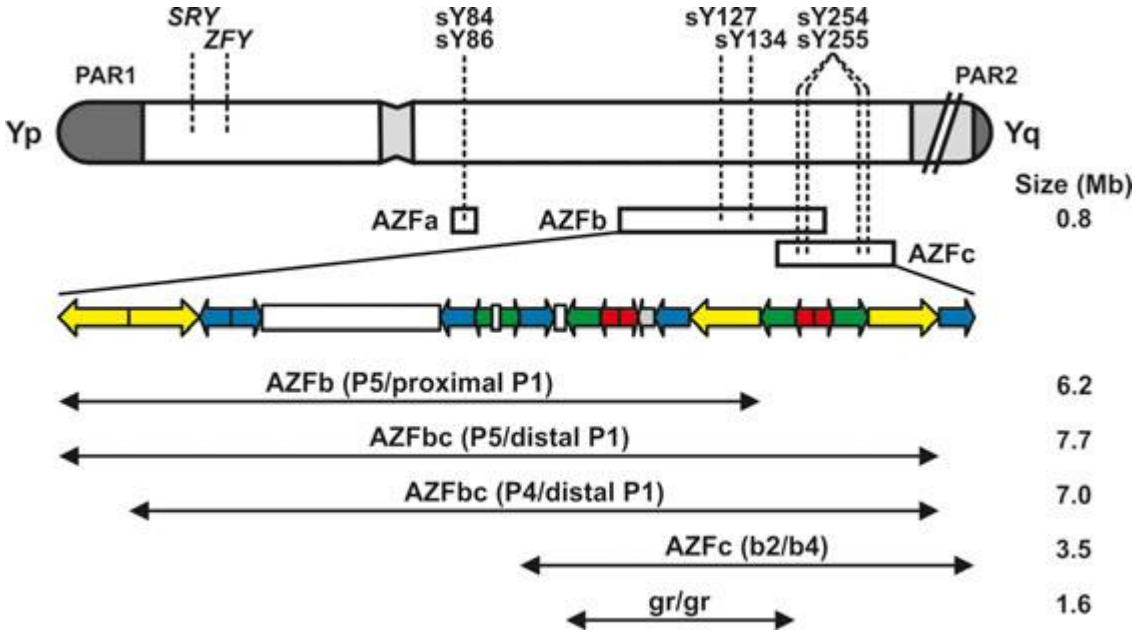
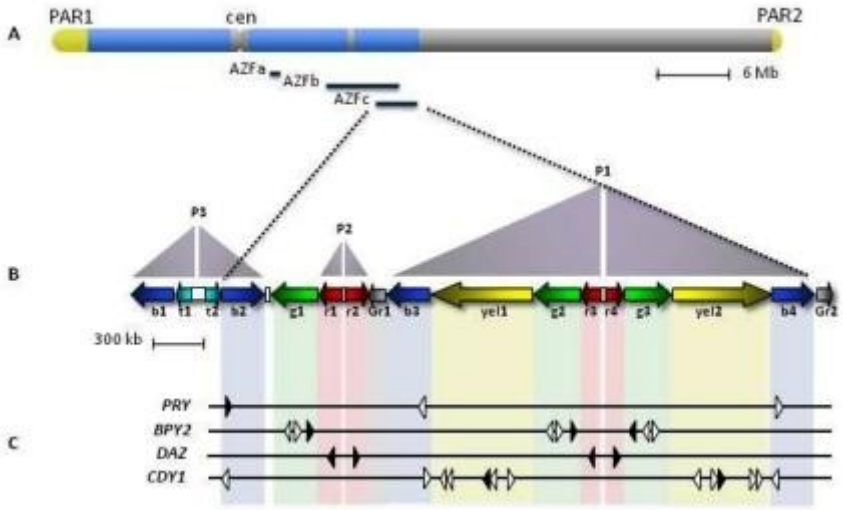


Figure 4 Schematic representation of the Y chromosome and the current microdeletion model (Repping et al., 2002). Repetitive sequences (colour coded palindromes) explain the origin of deletions in the AZFbc region by homologous recombination between identical sequences. The location of the STS primers suggested by the present guidelines is indicated by dashed lines. As four copies of the DAZ gene are normally present on the Y chromosome, the STS primers sY254, sY255 amplify four loci in AZFc. The AZFc (b2/b4) deletion is by far the most frequent type (~80%) of Y-chromosomal microdeletions found in men with severe oligo/azoospermia.

C. Krausz, L. Hoefsloot, M. Simoni and F. T€uttelmann "Andrology"

Andrology, 2014, 2, 5–19 *EAA/EMNQ best practice guidelines for molecular diagnosis of Y-chromosomal microdeletions: state-of-the-art 2013*



Figure– Genomic organization of the AZFc region of the Y chromosome. (A) Schematic view of the Y chromosome. The pseudoautosomal regions (PAR1 and PAR2) are depicted in light green and the euchromatic domains of the male-specific region of the Y in blue. The grey regions represent heterochromatic sequences (cen ¼ centromere). The three AZF regions are mapped below the ideogram. (B) Structural architecture of the reference AZFc region. AZFc is almost exclusively constituted by stretches of ampliconic units, each represented by block arrows. The AZFc amplicons are divided into five colour-coded sequence families (yellow, blue, green, red and grey) with each unit being designated according to a binomial notation indicative of family type and intra-family unit number. The length and orientation of the arrows represent amplicon size and polarity, respectively. Organization of amplicons in symmetrical arrays of contiguous repeat units (palindromes P1 to P3) is depicted by large triangles above the sequence. Single copy domains are indicated in white, although the spacers between the two red amplicon clusters are identical. (C) Mapping of the AZFc genes to the ampliconic units. Solid triangles represent active transcription units and white triangles depict pseudogenes. Orientation refers to 5′ –3′ polarity. Please note the high gene density of the interval and the extensive pseudogenic content.

Navarro-Costa et al., 2010 *Human Reproduction Update*,16(**5)**: 525-542. The AZFc region of the Y chromosome: at the crossroads between genetic diversity and male infertility.

A- Esquema do cromossoma Y, com os três domínios de regulação da espermatogénese (AZFa, AZFb e AZFc) indicados. A eucromatina está representada a azul e a heterocromatina a cinzento.

B- Arquitectura genómica da região AZFc. As setas coloridas indicam blocos de DNA repetitivo.

C- Conteúdo genético da região AZFc. Triângulos pretos representam cópias activas e triângulos brancos psedogenes.

Navarro-Costa et al., 2010 *Human Reproduction Update*,16(5): 525-542. The AZFc region of the Y chromosome: at the crossroads between genetic diversity and male infertility.

Table II - AZF microdeletions in infertile men

Pathological status	Karyotype	AZF microdeletions			SRY in X chr.
		AZFa	AZFb	AZFc	
Infertility	**46,XY**			**X**	
Azoospermic	**46,XY**	**X**			
Azoospermic	**46,XY**			**x**	
Azoospermic	**46,X,der(X)t(X;Y)(p22.31;p11.2)**	**x**	**x**	**x**	**X**
Azoospermic	**46,X,+mar,inv(5)/45,X,inv(5).ish i(Y)(p10)**	**x**	**x**	**x**	
Oligozospermic/OTA	**46,XY**			**x**	
Infertility	**46,XY**			**x**	
Cryptozoospermia	**46,XY**			**x**	
Azoospermic	**46,XY**			**x**	
Infertility	**46,XY**		**x**		
Infertility	**46,XY**			**x**	
Infertlity	**46,XY**			**x**	
Azoospermic	**46,X,der(X)t(X;Y)(p22.33;p11.3)**	**x**	**x**	**x**	**X**
Azoospermic	**46,XY**		**x**	**x**	
OTA	**46,XY**			**x**	
Azoospermic	**46,XY**			**x**	
Total cases 247	**16**	**4**	**5**	**14**	**2**
	6.5%	**1.6%**	**2.0%**	**5.7%**	

/

Table III – Number of Y microdeletions observed in 247 infertile men with azoospermia (AZO), oligozoospermia (OLIGO), oligo-asteno-teratozoospermia (OTA), astenotetozoospermia (AST), teratozoospermia (TER), cryptozoospermia (CRY) and infertile men with no indicated semen quality/status (NISQ), suspect of Klinefelter syndrome and "couple infertile" (CI) .

types	Number cases	% deleções	Del AZFa	Del AZFb	Del AZFc	Del grAZFc
Infertilidade (NISQ)				1	3	
Infertilidade Casal						
Azoospermia			4	4	8	1
Oligozoospermia						
Oligoteratoastenozoospermia-OTA					1	1
OT						
AO (OA						
Teratozoospermia						
Astenoteratozoospermia						
Criptozoospermia					1	
Hipogonadismo						
Total	**247**					

AZF microdeletions in infertile men

Pathological status	Karyotype	AZFa	AZFb	AZFc	gr AZFc	SRY absenc	SRY in X chr.
Infertility	46,XY			X			
Azoospermic	46,XY	X					
Azoospermic	46,XY			x			
Azoospermic	46,X,der(X)t(X;Y)(p22.31;p11.2)	x	x	x			X
Azoospermic	46,X,+mar,inv(5)/45,X,inv(5).ish i(Y)(p10)	x	x	x			
Oligozospermic/OTA	46,XY			x			
Infertility	46,XY			x			
Cryptozoospermia	46,XY			x			
Azoospermic	46,XY			x			
Infertility	46,XY		x				
Infertility	46,XY			x			
Infertlity	46,XY			x			
Azoospermic	46,X,der(X)t(X;Y)(p22.33;p11.3)	x	x	x			X
Azoospermic	46,XY		x	x			
OTA	46,XY				x		
Azoospermic	46,XY				x		
Total cases	**16 cases in 247**	4	5	12	2		2
	6.5%	1.6%	2.0%	4.9%	0.8%		

Meto as 2 gr dentro dos AZFc, pois não vou falar discutir elas à parte

Total deleções 23

AZF microdeletions in infertile men

Pathological status	Karyotype	AZF microdeletions				SRY absenc	SRY in X chr.
		AZFa	AZFb	AZFc	grAZFc		
Infertility	46,XY			X			
Azoospermic	46,XY	X					
Azoospermic	46,XY			x			
Azoospermic	46,X,der(X)t(X;Y)(p22.31;p11.2)	x	x	x			X
Azoospermic	46,X,+mar,inv(5)/45,X,inv(5).ish i(Y)(p10)	x	x	x			
Oligozospermic/OTA	46,XY			x			
Infertility	46,XY			x			
Cryptozoospermia	46,XY			x			
Azoospermic	46,XY			x			
Infertility	46,XY		x				
Infertility	46,XY			x			
Infertlity	46,XY			x			
Azoospermic	46,X,der(X)t(X;Y)(p22.33;p11.3)	x	x	x			X
Azoospermic	46,XY		x	x			
OTA	46,XY				x		
Azoospermic	46,XY				x		
Total cases	**16 cases in 247 samples**	4	5	12	2		2
	6.5%	1.6%	2.0%	4.9%	0.8%		

Total deleções 23

Meto as 2 gr dentro dos AZFc, pois não vou falar discutir elas à parte

Abnormal karyotypes and/or Y microdeletions

	2015 (até 30.03)				Estu Y	Del AZFa	Del AZFb	Del AZFc	S R Y
383373 háFoto	12.01	HGO CIRMA CAFert	infertilidade	46,XY,t(8;14) (p23.3;q11.2)	383374	-	-	-	
386857	27.01	H Lusíadas	Infertilidade	46,XY,t(1;10)	386858	-	-	-	
399891	05.03	HDE	Infertilidade masculina	45,XY,rob(13;14) (q10;q10)	399894	-	-	-	
400626	10.03	HGO CIRMA	Infertilidade	46,XY	400629	-	-	deleção	
405321	26.03	ARS Alentejo	Infertilidade	46,XY,t(4;22) (p10;q10)	421323	-	-	-	
406016	30.03	HDE	Infertilidade	47,XXY	416018	-	-	-	
24casos				5 anomalias = 20,8%		0	0	1	
	2014								
307991	21.01	HEgasMoniz	Azoospermia	47,XXY	N				
340571	20.06	HGO CIRMA CExInfertili	infertilidade	47,XY,+mar.ish der(14/22) (D14Z1/D22Z1+)	340574	-	-	-	
343092	03.07	Hgo	infertilidade	46,XY,t(1;3)(q42.3;q26.2)	N				
345958	17.07	HGO	infertilidade	47,XXY	345959	-	-	-	
349769	01.08	Chlc	azoospermia	47,XXY	349770	-	-	-	
368774	04.11	HGO	azoospermia	Mos 45,X[34]/46,X,idic(Y)(q11.221)[16]	N				
373887	26.11	HGO	azoospermia	46,XY	373890	deleçã	-	-	
376654	09.12	CHLC mac	azoospermia	46,XY	376655	-	-	deleção	
379364	18.12	MAC CHLC c.And	Infertilidade pri	47,XXY	379366	-	-	-	
380104	22.12	CHLC	Infertilidade	46,XY,inv(8)???	380103	-	-	-	
380592	29.12	CHLC	infertilidade	47,XXY	380594	-	-	-	
2014		65 casos	9 anomalias	13,8% anomalias		1		1	
	2013	REVISTO nossos c/ Y							
255601			*Susp S Klinefel-47,XXY NÃO ENTRA - 17 anos*		*N*				
270209 X	05.07	HGO tb pede FQ	Infertilidade	47,XXY	270211	-	-	-	
271413	11.07	HDE Genética	Estudo pré-concecional Hist familiar. Pede drepanocitoser	46,XY,inv(2)(p11.2q13) Não entra?	271414	Não i	o	vi	
293047	06.11	HGO	Infertilidade	47,XXY	293050	-	-	-	
		39 casos	3 anomalias =	7,7% anomalias		0	0	0	
	2012	Não fui ver outros "Não indicado" sem estudo Y	*Aqui estão todos de*	*Lista mercearia*	*B Molecular*				
175079	20.02	MAC	Azoospermia	46,XY	175080	-	-	deleção	
175993	24.02	CS ACES Oeste Norte	Infertilidade	47,XXY	175994	-	-	-	
182396 x	05.04	CHLC há FOTO	Infertilidad-OligoAcent	46,XY,t(4;8)(q27;q11.23)	182397	-	-	-	
188227	16.05	consultório	infertilidade	47,XXY	188228	-	-	-	
203679	14.08	Hévora	Infertilidade	46,XY,inv(14)	N				

x			**primária**						
	46,XY,	**Inv(14)(q13q22).ish**	**Inv(14)(p21.1)(RP11-388M7+)(q21.3)**	**(RP11-168D12+), *14q23.3(RP11-430G13x2)***					
Total	**2012**	**41 casos** (6 não entram)	**4 Cariot anormal**	**9,8% anomalias**	**Nº Y =**	**0**	**0**	**1**	
	2011								
164370 x	**15.12**	**MAC**	**Azoospermia**	**mos 47,XXY[41]/ 46,XY[09]**	N				
159998	**07.11**	**Particular - consultorio**	**Aazooospermia**	**47,XXY**	159999	-	-	-	
156217 x	**03.10**	**Arslvt lisboriental**	***Infertilidade primária*** *Tia com alter cromos.*	**46,XY,t(7;22)(q11.23;q11.2)**	156218	-	-	-	
146671 x	**14.07**	**ARSLVT**	**OTA oligoastenoteratozoos**	**mos 47,XXY[2]/46,XY[48]**	N				
142985	17.06	MAC	OTA	46,XY	142986			**gr/gr**	
138967 x	**19.05**	**MAC**	**Azoospermia**	**46,XY,dup(8)(p23.1p23.1). ish (wcp8+)**	138968	-	-	-	
137431 x	**10.05**	**HESanto Évora**	**Azoospermia**	**46,X,der(X) SRY está crom. X**	137434	**deleçã**	**deleçã**	**deleçã**	**No Cr. X**
		46,X,der(X)t(X;Y)(p22.31;p11.2).	ishder(X)t(X;Y)(p22.31;p11.2)(SRY+)						
137344 x	09.05	ARSLVTAcesLisboaNort	Desenv sex diminuido **NÃO ENTRA 47,XXY**		N				
136917 X	**05.05**	**MAC**	**Azoospermia**	**mos 47,XXY[13]/ 46,XX[37]**	136918	-	-	-	
133072	**08.04**	**MAC**	**Azoospermia**	**47,XXY**	133075	-	-	-	
133654	**13.04**	**ADSE**	**Azoospermia**	**46,X,+mar,inv(5)**	133657	**deleç**	**deleç**	**deleç**	
		46,X,+mar,inv(5)	(p14.2p15.2)[32]/ 45,X,inv(5)(p14.2 p15.2)[18]. Ish i(Y)	(p10)(SRY++,DYZ3+),inv(5)(wcp5+)					
130636	**24.03**	**MAC**	**Azoospermia**	**47,XXY**	130637	-	-	-	
129174 X	15.03	MAC	OligozoosperAcentu OTA	46,XY	129175	-	-	**deleção**	
127384	01.03	consultorio	infertilidade		127387	-	-	**deleção**	
125161	14.02	MAC	OTA	46,XY	125162	-	-	-	
125376 X	**15.02**	**MAC**	**Oligozoospermia**	**46,XY,rob(13;14)+mar**	N				
			46,XY,rob(13;14)+mar.	**Ish der(16)(wcp16+)**					
119484	13.01	MAC	criptozoospermia	46,XY	119486	-	-	**deleção**	
118775 X	**11.01**	**MAC**	**OTA**	**46,XY,t(4;14;15)**	N				
46,XY,	T(4;14;15)	(q22;q21;q21.39??).	Ish der(4)t(4;15)(wcp15+)	,der(14)t(4;14)(wcp4+),der(15)t(14;15)					
118779 X	**11.01**	**MAC**	***Infer, 38 anos*** *baixa estatura*	**46,X,+mar.ish der(Y) (wcpY+,DYZ3+)**	N				
Em	2011	**106 /104 casos** (4 não entram)	**13 anomalias** (1 não entra)	**12,3% anomalias**	**Y = %**	**2**	**2**	**5**	
								gr/gr	

	2010								
60379	15.01	MAC	Azoosperm secretória	46,XY	60380	-	-	**deleção**	
62688	27.01	ARSLVT Oeste Sul	AR não entra 46,xy **Azoospermia**	46,XY	62689	-	**deleçã**	**deleção**	
65148	**11.02**	**ARSLVTACESOesteNort**	**Infertilidade**	**46,XY,rob(13;14)(q10;q10)**	N				
66641 X	23.02	TiagoR e CS St Isabel CEMEARE	Hipogonadismo a esclarecer Suspeita Klinefelter? **Nãoentra? 42anos**	*Mos 47;XXY[48]/46,XY[2]*	N				
68520	**05.03**	**MAC**	**Azoospermia por hipogonadismo primário**	**47,XXY**	**N**				
70910 X	*19.03*	*ARSLVT ACESLisboaNort*	*Esposa 2AE Não entra*	*46,XY,inv(1)(p13q12) variante*	*N*				
74242 x	09.04	Consultório	Suspeita S Klinefelter Não conta? 41 anos	*46,X,der(X)t(X;Y(p22.33;p11.3)*	N				
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33;;p11.3). ish der(X)t(X;Y)	(p22.33;p11.3)(SRY+)					
74900	14.04	MAC	infertilidade	46,xy	74901	-	**deleçã**	-	
76981		MAC	infertilidade	46,XY	76982	-	-	**deleção**	
79446 X	**10.05**	**MAC**	**Azoospermia secretória**	**der(X)t(X;Y(p22.33;p11.3) (wcp**	79447	**deleçã**	**deleçã**	**deleção**	**no X**
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33).ishder(X)t(X;Y)	(p22.33;p11.3)(wcpY+;SRY+)					
80750	*19.05*	*MAC Andrologia*	*Hipogonadismo primário* *Susp S. Klinefelter*	*47,XXY*	*N*				
84664	**14.06**	**MAC**	**Criptozoospermia**	**47,XXY**	84668	-	-	-	
91268	**12.07**	**MAC**	**Azoospermia. Hipogonadismo primário**	**47,XXY**	91269	-	-	-	
95875	**12.08**	**MAC**	**Azoospermia**	**47,XXY**	N				
101373	**17.09**	**MAC**	**Azoospermi secretória**	**47,XXY**	101376	-	-	-	
116498 X	**27.12**	**MAC**	**OTA**	**47,XYY**	N				
Se não entrar com	**2015**	**Devido desvirtuar do HGO não indicados**	**Perco 4 translocações e XXY**						
383373	12.01	HGO	Não indicado	46,XY,t(8;14)	383374				
386857+	27.01	H Lusíadas	Infertilidade	46,XY,t(1;10)	386858				
399891+	05.03	HDE	Infertilidade	45,rob(13;14)(q10;q10)	399894				
405321+	26.03	ARS Alentejo	Infertilidade	46,XY,t(4;22)(p10;q10)	421323				
406016+	30.03	HDE	Infertilidade	47,XXY	416018				

Chromosomal disorders and male infertility

Simão L[1]; Brito F[1]; Gonçalves J[2]; Caetano I[2]; Marques B[1]; Pedro S[1]; Alves C[1]; Silva M[1]; Serafim S[1]; Ambrósio P[1]; Geraldes MC[1];Correia H[1].

[1]Unidade de Citogenética, Departamento de Genética Humana, Instituto Nacional de Saúde Dr. Ricardo Jorge, INSA,IP, Lisboa. [2]Unidade de Genética Molecular, Departamento de Genética Humana, Instituto Nacional de Saúde Dr. Ricardo Jorge, INSA,IP, Lisboa.

INTRODUCTION

Infertility is defined as the inability to conceive after one or more years of unproctected intercourse (Shah,2003) and occurs in approximately 15% of the population (Goel,2010; Harton,2012), affecting about 7% of men (Forti,1998). Male factor infertility is considered a complex disorder with a largely unknown etiology (O'Brien,2010, Hotaling,2014). In general, genetic abnormalities are thought to account for 15%-30% of this condition (Ferlin, 2007) and Y chromosome microdeletions are also frequently implied (Galletto,2008; O`Flynn;2010).

The study, based on our casuistic, aimed at contributing to a better understanding of the genetic causes of infertility, in order to improve genetic counseling of these conditions.

MATERIAL AND METHODS

A group of 410 idiopathic infertile men with non-obstructive azoospermia, oligozoospermia, or unknown semen quality (based on clinical evaluation and/or sperm counts) was retrospectively selected. Conventional karyotype was performed in all samples; Y chromosome microdeletion testing/study was performed in 247 samples.

RESULTS AND DISCUSSION

Forty two abnormal karyotypes (10.2%) were found, indicating an elevated frequency of chromosome abnormalities among the selected infertile men, as compared to that of newborn populations (≈0.4%, Van Assche,1996). This frequency is higher than that reported in similar studies that pointed to frequencies ranging from 2.2%-14.3% (Gekas,2001).

As expected, Klinefelter´s syndrome was the most common chromosome disorder (4.9%). There were 18 cases with 47,XXY karyotype and 2 cases of mosaicism involving lines 47,XXY and 46,XY. Reciprocal translocations were identified in 10 cases (2.4%) and, particulary, in men with unknown semen quality. These results agree with finding from other studies reporting approximately 1% of infertile men with this type of anomalie (Harton,2012;Goel,2010) but differ in that no association was detected with oligozoospermia (Van Assche,1996). On the other hand, Y microdeletions were identified in 16/247 cases (6.5%), more frequently in azoospermics (11.7%, corresponding to 7/60 azoospermics), of whom 6/ 7 presented deletions at the AZFc region.
The marked presence of chromosomal abnormalities and Y chromosome microdeletions enphasizes the relevance of studying both factors in infertile men to improve genetic counseling, to allow the development of appropriate therapies, and to expand the knowledge about the ethiology of male infertility.

These results reforce the importance of cytogenetic and molecular studies in the abordage of infertile men population

Chromosomal disorders and male infertility

Simão L; Brito F; Gonçalves J; Caetano I; Marques B; Pedro S; Alves C; Silva M; Serafim S; Ambrósio P; Geraldes MC;Correia H.

INTRODUCTION

Infertility is defined as the inability to conceive after one or more years of unproctected intercourse (Shah,2003) and is common occurring in approximately 15% of the population (Goel,2010;Harton,2012).
Infertility affects about 7% of all men (Forti,1998). So, male factor infertility is a complex disorder that affects a large sector of the population; however, many of its etiologies are unknown (O'Brien,2010,Hotaling,2014). Genetic abnormalities are thought to account for 15%-30% of male factor infertility (Ferlin, 2007). This study was conducted in order to contribute to a better understanding of the genetic causes of infertility and to be able to provide proper genetic counseling.

MATERIAL AND METHODS

We retrospectively evaluated the results of a group of 410 idiopathic infertile men with non-obstructive azoospermia, oligozoospermia, as well as infertile men with unknowed semen quality. Subjects were selected based on clinical evaluation and/or on sperm counts.
We will focus on our current understanding of the chromosomal basis of male infertility specifically: numerical or structural karyotype abnormalities and Y chromosomal microdeletions.
Conventional karyotype was performed in all the samples. Y chromosome microdeletion testing/study were performed in **247** samples.

RESULTS AND DISCUSSION

Forty two abnormal karyotypes were found corresponding to a frequency of **10.2**%. These results indicates an elevated frequency of chromosome abnormalities among the infertile men as compared to that of newborn populations (≈0.4%, Van Assche,1996). This value is relatively elevated when compared to the results of similar studies, which have shown frequencies of chromosomes abnormalities ranging from 2.2%-14.3% (Gekas,2001).
As expected, Klinefelter´s syndrome was the most common cytogenetic anomaly. There are 18 cases with 47,XXY karyotype and 2 cases of mosaicism involving lines 47,XXY and 46,XY (**4,9**% of total sample).
Reciprocal translocations are present in **10** cases **(2,4%)**, particulary in the group of infertile men with unknowed pathological semen quality and this structural anomalies seem to contribute to infertility (Harton,2012;Goel,2010). A review study reveals that reciprocal translocations have been found in approximately 1% of the infertile men and are more common in azoospermic than in oligozoospermic males (Van Assche,1996), a relation not found in the present study.
Y chromosome microdeletions are a frequent cause of infertility in males (Galletto,2008; O`Flynn;2010). Y microdeletions were identified in **16** of the **247** Y microdeletions cases (**6.5**%). There was more frequent in azoospermics, **7** cases of the idenfied 60 azoospermic males (11.7%). Most azoospermic cases (**6 in 7**) presented deletions in AZFc region.
The significative presence of chromosomal abnormalities and Y chromosome microdeletions enphatisize the study of both factors in infertile men as an essential stage for the development of appropriate therapies, genetic counseling and in the expansion of the knowledge of the masculine infertility.

These results reforce the importance of cytogenetic and molecular studies in the abordage of infertile men population

Despite major advances in the diagnostic workup of infertile males, the etiopathogenesis of testicular failure remains undefined in about 50% of cases and are referred to as “idiopathic infertility”(Krausz,2011).

Quadro geral nº anomalias (1 é existência de deleção)

Ano	Amostras	Anomi	Anom numé	Anom estrut	% anomi	Casos Y deleçõ	AZFa	AZFb	AZFc	Gr AZFc	gra?	SRY
2015	**23**	**5**	1	4	**21,7**	400629 Infertilidade	-	-	1 Infer			
2014	**64**	**9**	6	3	**14,1**	373890 Azoospermia	1 Azoo	-	-			
						376655 azoospermia	-	-	1 Azoo			
2013	**38**	**3**	2	1	**7.9**	--------						
2012	**38**	**4**	2	2	**9.8**	175080 Azoospermia	-	-	1 Azoo			
2011	**106**	**13**	6	7	**12.3**	142986 OTA				1 OTA		
						137434 Azoosp der(X)t(X;Y)	1	1	1 Azoo			**Está X**
						133657 Azoosp mos 45,X/46 +mar i(Y)(p10)	1	1	1 Azoo			
						129175 Oligozoosper	-	-	1 Olig			
						127387 Infertilidade	-	-	1 infer			
						119486 Cryptozoosp	-	-	1 crypt			
							2	2	5			X
2010	**141**	**9**	6	2	**6.4**	60380 Azoospermia	-	-	1 azoo			
						62689 azoospermia	-	1 azo	1 Azo			
						68667 Infertilidade	-	-	1 Infer			
						74901 Infertilidade	-	1 infer	-			
						76982 infertilidade	-	-	1 Infer			
						79447 Azoospermia Der(X)t(X;Y)	1 azoo	1 azoo	1 azoo			Está **X**
							1	3	5	1gr	1?	x
Total	**410**	**43**	23	19	**10,2**	Total deleções 23 ou 24 (gr?)	4 2.5%	5 3.1%	13 8.1%	1	?	2?
						Em 16 casos - 9.9%						

groups

Group	15	14	13	12	11	10	No. Individuals	Sex chr No.	Sex chr %	Autosomes No.	Autosomes %	Total chr. abnormal No.	Total %
AZO	4	16	1	6	26	32	**85**						
OLIGO			1	5	9	2	**17**						
OTA		1	2	10	27	43	**83**						
OT					2	2	**4**						
OA					1	1	**2**						
AST					1	12	**13**						
TER			1		6	9	**16**						
CRYP					4	4	**8**						
ICSI													
Infer NISQ	13	35	19	16	26	29	**138**						
Infer Couple	4	10	14	1	4	7	**40**						
Não indi	2	2					**4**						
Total **410**	**23**	**64**	**38**	**38**	**106**	**140**	**410**					**44**	**10.6**

Tirei os 3 questionaveis

Y chromosome microdeletions are a frequent cause of infertility in males (Galletto,2008;Gonçalves,1997). Y microdeletions were identified in **16** of the **161** Y microdeletions cases/samples analysed (**9,9**%), more significantly in azoospermics (**7** cases (____% of the ____azoospermics studed). In same of them (**3** azoospermic cases), deletions in differents AZF regions are present (total **19** deletions). Most azoospermic cases (**6 in 7**) presented deletions in AZFc region.
The XY bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome… (Thomas,2000) We found **1** case with karyotype 46,X,der(X)t(X;Y). The SRY is localizated on X chromosome.

Chromosomal translocations may cause reductions in testicular volume and testosterone level, which may impact spermatogenesis, resulting in aazoospermia or oligozoospermia and male infertility (Dong,2012) Reciprocal translocations are present in **10 cases (2,4%)**, particulary in the group of infertile men with no indicated semen quality (NISQ unknowed pathological semen quality. In fact, _____% of this individuals have a structural rearrangement, and this fact seems to contribute to infertility, salientando importancia destas anomalias cromossómicas in male infertility (Harton,2012;Goel,2010). A review study reveals that reciprocal translocations have been found in approximately 1% of the infertile men and are more common in azoospermic than in oligozoospermic males (Van Assche,1996), a relation not found in the present study.
Y chromosome microdeletions are a frequent cause of infertility in males (_____,_____). Y microdeletions were identified in 13 cases of the microdeletions 161 samples analysed (8.1%), more significantly in azoospermics (7 cases (____% of the azoospermics). In same of them (3 azoospermic cases), deletions in differents AZF regions are present (total 19 deletions). Most azoospermic cases (6 in 7) presented deletions in AZFc region..
The XY bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome… (Thomas, 2000) We found three cases with karyotype 46,X,der(X)t(X;Y). in all of those the SRY is localizated on X chromosome.

Quadro geral nº anomalias Y (1 é existência de deleção)

Ano	Amostras	Anomi	Anom numé	Anom estrut	% anomi	Casos Y deleçõ	AZF a	AZFb	AZFc	Gr AZF c	gr a?	SRY
2015	**24**	**5**	1	4	**20.8**	400629 Infertilidade	-	-	1 Infer			
2014	**65**	**9**	6	3	**13.8**	373890 Azoospermia	1 Azo	-	-			
						376655 azoospermia	-	-	1 Azoo			
2013	**38**	**3**	2	1	**7.9**	--------						
2012	**38**	**4**	2	2	**9.8**	175080 Azoospermia	-	-	1 Azoo			
2011	**106**	**13**	6	7	**12.3**	142986 OTA				1 OTA		
						137434 Azoosp der(X)t(X;Y)	1	1	1 Azoo			**Está X**
						133657 Azoosp mos 45,X/46 +mar i(Y)(p10)	1	1	1 Azoo			
						129175 Oligozoosper	-	-	1 Olig			
						127387 Infertilidade	-	-	1 infer			
						119486 Cryptozoosp	-	-	1 crypt			
							2	2	5			X
2010	**144** **141**	**11** **8**	8 6	3 2	**7,6** **5,7**	60380 Azoospermia	-	-	1 azoo			
						62689 azoospermia	-	1 azo	1 Azo			
						68667 Infertilidade	-	-	1 Infer			
						74901 Infertilidade	-	1 infer	-			
						76982 infertilidade	-	-	1 Infer			
						79447 Azoospermia der(X)t(X;Y)	1 azoo	1 azoo	1 azoo			Ausente Y **VER**
							1	3	5	2gr	1?	x
Total	**410**				**10,2**	**Total deleções 25?**	4 2.5%	5 3.1%	13 8.1%	2	?	2?
						Em 16 casos -6.5%						

N and %

	N cases	% cases
Total Karyotypes studis	**410**	
Normal Karyotypes	368	
Abnormal karyotype	42	10.7
Sex chromosomes	28	6.8
autossomes	16	3.9
Total Y microdeletions studies	247	
Normal Y microdeletions studies	231	
Y microdeletions	16	8.7
Abnormal karyotype with Y microdeletions		

Y chromosome microdeletions are a frequent cause of infertility in males (_____,_____). Y microdeletions were identified in 13 cases of the microdeletions 161 samples analysed (8.1%), more significantly in azoospermics (7 cases (____% of the azoospermics). In same of them (3 azoospermic cases), deletions in differents AZF regions are present (total 19 deletions). Most azoospermic cases (6 in 7) presented deletions in AZFc region..

The XY bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome… (Thomas, 2000). We found two cases with karyotype 46,X,der(X)t(X;Y) with the assence of SRY in chromosome Y / SRY localizated on X chromosome. CONFIRMAR

AZF microdeletions in infertile men

Pathological status in Different cases	Karyotype	AZFa	AZFb	AZFc	gr AZFc	SRY absenc	SRY in X chr.	Case number
Infertility	46,XY			X				400626/ 400629
Azoospermic	46,XY	X						373890
Azoospermic	46,XY			x				175080
Azoospermic	46,X,der(X)t(X;Y)(p22.31;p11.2) 137431	x	x	x			X	137434
Azoospermic	46,X,+mar,inv(5)/45,X,inv(5).ish i(Y)(p10)	x	x	x				133657
Oligozospermic/OTA	46,XY			x				129175
Infertility	46,XY			x				127387
Cryptozoospermia	46,XY			x				119486
Azoospermic	46,XY			x				60380
Infertility	46,XY		x					74901
Infertility	46,XY			x				68667
Infertlity	46,XY			x				76982
Azoospermic	46,X,der(X)t(X;Y)(p22.33;p11.3)	x	x	x			X	79447
Azoospermic / ARepetição	46,XY		x	x				62689
OTA	46,XY				x			142986
Azoospermic	46,XY				x			376655
Total cases	**16 cases in 247**	4	5	12	2		2	
	6.5%	1.6%	2.0%	4.9%	0.8%			

Total deleções 23

AZF microdeletions in infertile men

Pathological status in Different cases	Karyotype	AZFa	AZFb	AZFc	SRY in X chr.	Case number

Infertility	**46,XY**			**X**		400626/ 400629
Azoospermic	**46,XY**	**X**				373890
Azoospermic	**46,XY**			**x**		175080
Azoospermic	**46,X,der(X)t(X;Y)(p22.31;p11.2) 137431**	**x**	**x**	**x**	**X**	137434
Azoospermic	**46,X,+mar,inv(5)/45,X,inv(5).ish i(Y)(p10)**	**x**	**x**	**x**		133657
Oligozospermic/OTA	**46,XY**			**x**		129175
Infertility	**46,XY**			**x**		127387
Cryptozoospermia	**46,XY**			**x**		119486
Azoospermic	**46,XY**			**x**		60380
Infertility	**46,XY**		**x**			74901
Infertility	**46,XY**			**x**		68667
Infertlity	**46,XY**			**x**		76982
Azoospermic	**46,X,der(X)t(X;Y)(p22.33;p11.3)**	**x**	**x**	**x**	**X**	79447
Azoospermic	**46,XY**		**x**	**x**		62689
OTA	**46,XY**			**X**		142986
Azoospermic	**46,XY**			**x**		376655
Total cases	**16 cases in 247**	**4**	**5**	**12**	**2**	
	6.5%	**1.6%**	**2.0%**	**5.7%**		

Table III – Number and frequency of chromosome abnormalities observed in ____ infertile men with azoospermia (AZO), oligozoospermia (OLIGO), oligo-asteno-teratozoospermia (OTA), astenotetozoospermia (AST), teratozoospermia (TER), cryptozoospermia (CRY) and infertile men with no indicated semen quality/status (InferNIQ).

Group	No. individuals	Sex chr No.	Sex chr %	Autosomes No.	Autosomes %	Total chr. abnormal No.	Total %
AZO							
OLIGO							
OTA							
AST							
TER							
CRY							
Infer NIQ							
CI							
Total	**410**					**42**	**10.2**

Chromosomal disorders and male infertility

Simão,L.;Brito,F.;Gonçalves,J.;Caetano,I.;Marques,B.;Alves,C.;Silva,M; Serafim,S; Ambrósio,P; Geraldes,MC;Correia,H.

Médicos são MAC- CHLC, HGO, HDE, mas muitos + ACES, etc não pôr nenhum? Ou só os 2 sítios com mais amostras , quem é no HGO, quem é na MAC-CHLC?

INTRODUCTION

Infertility is defined as the inability to conceive after one or more years of unproctected intercourse (Shah,2003). Infertility in humans is common occurring in approximately 15% of the population wishing to start a family (Goel,2010;Harton,2012). Despite this, the molecular and genetic factors underlying the cause of infertility remain largely undiscovered(Harton,2012) **INTERESSE DO TRABALHO**(_____).
Infertility affects about 7% of all men (Forti,1998)**Male factor É responsável**
Male factor infertility is a complex disorder that affects a large sector of the population; however, many of its etiologies are unknown (O'Brien,2010, Hotaling,2014). Genetic abnormalities are thought to account for 15%-30% of male factor infertility (Ferlin, 2007). This study was conducted in order to contribute to a better understanding of the genetic causes of infertility and to be able to provide proper genetic counseling. **INTERESSE DO TRABALHO**

MATERIAL AND METHODS

We retrospectively evaluated the results of a group of idiopathic infertile men with non-obstructive azoospermia, oligozoospermia, as well as infertile men with unknowed semen quality. The studies were conducted in 415 blood samples from infertile males.
Subjects were selected based on clinical evaluation and/or on sperm counts.
We will focus on our current understanding of the chromosomal basis of male infertility specifically: chromosomal aneuploidy, structural or numerical karyotype abnormalities and Y chromosomal microdeletions.
Conventional karyotype was performed in all the samples. Fluorescence *in situ* hybridization (FISH) studies were performed when necessary/justified. Y chromosome microdeletion testing/studies were performed in 161 samples.

RESULTS Focar mais a CITOGENÉTICA

Forty four abnormal karyotypes were found corresponding to a frequency of 10.6%. This results indicates/shows an elevated frequency of chromosome abnormalities among the infertile men as compared to that of newborn populations (≈0.4%, Van Assche,1996). This value is also relatively elevated when compared to the results of similar studies, which have shown frequencies of chromosomes abnormalities ranging from 2.2%-14.3% (Gekas,2001).

As expected, Klinefelter´s syndrome was the most common cytogenetic anomaly. There are 19 47,XXY karyotypes and 3 cases of mosaicism involving lines 47,XXY and 46,XY **(5,3% of total samples**).
Chromosomal translocations may cause reductions in testicular volume and testosterone level, which may impact spermatogenesis, resulting in aazoospermia or oligozoospermia and male infertility (Dong,2012) Reciprocal translocations are present in **10 cases (2,4%)**, particulary in the group of infertile men with no indicated semen quality (NISQ unknowed pathological semen quality. In fact, _____% of this individuals have a structural rearrangement, and this fact seems to contribute to infertility, salientando importancia destas anomalias cromossómicas in male infertility (Harton,2012;Goel,2010). A review study reveals that reciprocal translocations have been found in approximately 1% of the infertile men and are more common in azoospermic than in oligozoospermic males (Van Assche,1996), a relation not found in the present study.
Y chromosome microdeletions are a frequent cause of infertility in males (_____,_____). Y microdeletions were identified in 13 cases of the microdeletions 161 samples analysed (8.1%), more significantly in azoospermics (7 cases (____% of the azoospermics). In same of them (3

azoospermic cases), deletions in differents AZF regions are present (total 19 deletions). Most azoospermic cases (6 in 7) presented deletions in AZFc region..
The XY bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome… (Thomas, 2000) We found three cases with karyotype 46,X,der(X)t(X;Y). in all of those the SRY is localizated on X chromosome.

As expected, Klinefelter´s syndrome was the most common cytogenetic anomaly. There are 19 47,XXY karyotypes and 3 cases of mosaicism involving lines 47,XXY and 46,XY **(5,3% of total samples**).

Chromosomal translocations may cause reductions in testicular volume and testosterone level, which may impact spermatogenesis, resulting in aazoospermia or oligozoospermia and male infertility (Dong,2012) Reciprocal translocations are present in **10 cases (2,4%)**, particulary in the group of infertile men with no indicated semen quality (NISQ unknowed pathological semen quality. In fact, _____% of this individuals have a structural rearrangement, and this fact seems to contribute to infertility, salientando importancia destas anomalias cromossómicas in male infertility (Harton,2012;Goel,2010). A review study reveals that reciprocal translocations have been found in approximately 1% of the infertile men and are more common in azoospermic than in oligozoospermic males (Van Assche,1996), a relation not found in the present study.

DISCUSSION

The elevated frequency of chromosome abnormalities found in infertile men and particulary in the azoospermics (_____% of which has a chromosomal abnormality) emphasizes the role of these studies towards the understanding of the basis of male infertility. These studies and the complementary molecular analysis allow the provision of genetic counseling to a significant proportion of couples with male infertility factors.

The presence of sSMC maybe one of basis of men infertility because of interference of additional chromosome in meiotic process(Gardner, 2012).

As translocações reciprocas e aparentemente equilibradas, além de estarem relacionadas com infertilidade e perdas fetais em caso de gravidez, comportam para os individuos portadores um risco consideravel de t(GARner, 2004). Dado que o doente veio referenciado por oligozoospermia acentuada não é de excluir a correlação entre a indicação clinica e a alteração cros observada

astenotetozoospermia (AST), teratozoospermia (TER), cryptozoospermia (CRY) and infertile men with no indicated semen quality (Infer NIS), suspect of Klinefelter syndrome and “couple infertile” (CI).

REFERIR (JG) Importância da informação clínica completa e precisa (sobre a existência de dados da qualidade semen por exemplo , para se poder comparar) . Prevenção da doença genética na população

Despite major advances in the diagnostic workup of infertile males, the etiopathogenesis of testicular failure remains undefined in about 50% of cases and are referred to as “idiopathic infertility”(Krausz,2011).

Novas metodologias

Só infertilidade não tem informação

References
-Shah et al, 2003
-Harton and Tempest, 2012
-Goel et al, 2010; -Krausz et al, 2011

2001

Infertility affects 13-15% of the couples in western societies, with approximately half of the cases due to male factors[1,2,3]. Among the latter, the primary cause is deficient spermatogenesis revealed by abnormal sperm count and motility (oligozoospermia, azoospermia, teratozoospermia and astenozoospermia). On the other hand, abnormal spermatogenesis can be due to several factors, of which chromosomal aberrations are among the most important[4,5]. It is estimated that 20% of male infertility can be explained by abnormalities in mitotic and/or meiotic chromosomes[2].
The outlook for patients with severe male infertility has improved since the development of assisted reproductive technologies (such as intracytoplasmic sperm injection)[6]. However, based on the above data, chromosome analysis is widely recommended for the male partner undergoing medically assisted reproduction[5].
In the present study we investigated cytogenetically a group of idiopathic infertile men with non-obstructive azoospermia and oligozoospermia, in order to contribute to a better understanding of the genetic causes of infertility and to be able to provide proper genetic counselling.

METHODS

Cytogenetic studies were conducted in 349 blood samples from infertile males candidates for Medically Assisted Reproduction Programs showing azoospermia (91) or oligozoospermia (258). Subjects were selected based on clinical evaluation and on sperm counts*.
Chromosome analysis was performed on GTL-banded metaphases obtained from lymphocyte cultures of peripheral blood according to Yunis *et al* (1978) modified. Ag-NOR banding, C-banding and DAPI staining were performed when necessary, by standard procedures.
Fluorescence *in situ* hybridization (FISH) analysis was performed using: whole chromosome painting probes (wcp) for chromosomes X and Y (Cambio); centromeric-specific DNA probes for chromosomes 15 (pMC15), 21 (pZ21A), X (pBAMX5) and Y (pDP97); probe for pseudoautosomic region X/Y (pDP230); probes for the regions Yp11.2 (GMGY10), Yq11.23 (49f), Yq12 (pYH2.1) and 21q22.2-22.3 (D21S65) (Oncor).
Probes were labelled with biotin-14-dATP and digoxigenin-11-dUTP by nick translation. Hybridization, posthybridization washes and detection were performed according to Vieira *et al* (1999).

*Defined according to World Health Organization (WHO) recommendations and standards[7]

RESULTS

The results of the cytogenetic analysis of the 349 infertile men are presented in Table I. Forty abnormal karyotypes were found (34 in the azoospermic group and 6 in the oligozoospermic), corresponding to a frequency of 11.5%. This result indicates an elevated frequency of chromosome abnormalities among the infertile men as compared to that of newborn populations (~0.4%)[2]. That value is also relatively elevated when compared to the results of similar studies, which have shown frequencies of chromosome abnormalities ranging from 2.2%-14.3%[10], with an estimated overall frequency of 7.1%[11].
Chromosome abnormalities were by far more frequent in the azoospermic group (37.4%) than in the oligozoospermic (2.3%). These data seem to confirm a relationship between declining sperm counts and increasing frequency of chromosome abnormalities[2]. However, our results also indicate a higher frequency of chromosome abnormalities among the azoospermics than that reported by others (14.1%-

21.6%)[11,12] .The discrepancies may be due to differences in sample sizes analysed and/or the kind of selection applied to the infertile group.

Sex chromosome aberrations were highly predominant in the azoospermic group (97%), the majority of which (91%) are X chromosome abnormalities (Table II). It is known that these chromosomal disorders can interfere with testicular differentiation or maturation, as seen in numerical disorders of the X chromosome and in many structural and numerical disorders of the Y chromosome[13]. Our results show a high incidence of mosaics or full 47,XXY karyotypes. Indeed, Klinefelter's syndrome represents one of the most common causes of azoospermia[10].

(1) Moosani, N. *et al* (1995) *Fertil. Steril.*, **64**, 811-817.
(2) Van Assche, E. *et al* (1996) *Hum. Reprod.*, **11** (Suppl. 4), 1-24.
(3) De Kretser, D. *et al* (1999) *J. Clin. Endocrinol. Metab.*, **84**, 3443-3454.
(4) Thielemans, B. *et al* (1998) *Eur. J. Obst. Gynec.*, **81**, 217-225.
(5) De Braekeleer, M. and Dao, T.-N.(1991) *Hum. Reprod.*, **6**, 245-250.
(6) Palermo, G. *et al* (1992) *Lancet*, **340**, 17-18.
(7) World Health Organization (1993) *WHO manual for the standardized investigation and diagnosis of the infertile couple.* Cambridge University Press, New York.
(8) Yunis, J. *et al* (1978) *Chromosoma* **67**, 293-297.
(9) Vieira, L. *et al* (1999) *Cancer Genet. and Cytogenet.*, **114**, 17-21.
(10) Gekas, J. *et al* (2001) *Hum. Reprod.*, **16**, 82-90.
(11) Retief, A. *et al* (1984) *Hum. Genet.*, **66**, 162-164.
(12) Kjessler, B. *et al* (1966) *Monographs in Human Genetics*. vol. **2**. S. Karger, Basel.
(13) Bonaccorsi, A. *et al* (1997) *Fertil. Steril.*, **67**, 928-931.

2015

Infertility is defined as the inability to conceive after one or more years of unproctected intercourse (Shah,2003). Infertility in humans is common occurring in approximately 15% of the population wishing to start a family (Harton,2012). Despite this, the molecular and genetic factors underlying the cause of infertility remain largely undiscovered (_____). Male factor infertility is a complex disorder that affects a large sector of the population; however, many of its etiologies are unknown(O'Brien,2010).

MATERIAL AND METHODS

COM INDICAÇÂO DE INFERTILIDADE MASCULINA E INFERTILIDADE do CASAL(AR AE, Hipogo 15 anos não contam)

We retrospectively evaluated the results of a group of *idiopathic infertile men* infertile men with non-obstructive azoospermia, oligozoospermia and other sperm pathologies, as well as infertile men with unknowed pathological semen quality.

Subjects were selected based on clinical evaluation and/or on sperm counts.The study was conducted in order to contribute to a better understanding of the genetic causes of infertility and to be able to provide proper genetic counseling.

We will focus on our current understanding of the chromosomal basis of male infertility specifically: chromosomal aneuploidy, structural or numerical karyotype abnormalities and Y chromosomal microdeletions.

The studies were conducted in 414 blood samples from infertile males. Following karyotyping, fluorescence *in situ* hybridization (FISH) were performed when **necessário**. Y chromosome microdeletions are a frequent cause of infertility in males and Y chromosome microdeletion testingstudies were performed in 161 samples.

FISH 70910 sondas centroméricas, wcp1, P1.77heterocromática

RESULTS Focar mais a CITOGENÉTICA

Reciprocal balanced translocations are present in 9 cases inversions in 4 cases, and this fact seems to contribute to infertility, particulary in the group with unknowed pathological semen quality (___%). In this group ____% of individuals have a structural rearrangement, salientando importancia destas anomalias cromossómicas in male infertility (Harton,2012; Goel,2010).

Y microfdeletions in 3,1% of the cases, more significantly in azoospermics (____% of the azoospermics.

The XY bivalent is particularly susceptible to errors in meiosis because of homology between the X and the Y chromosome… (Thomas, 2000)

Anos

Year	NCases	AbnorKaryo	% abnormal	NCases	AZFa	AZFb	AZFc	Gr/grAZFc	SRY absence
2015	23	5	20.8	KA	0	0	1		
2014	64	9	13.8	L	1	0	1		
2013	39	3	7.7	M	0	0	0		
2012	38	4	10.5	N	0	0	1		
2011	106	12	11.3	O	2	2	5	1	
2010	1401	11	7.8	P				1	
Total	**410**	**42**	**10.2**						

Anos

Year	NCases	AbnorKaryo	% abnor	NCases	AZFa	AZFb	AZFc	Gr/grAZFc	SRY in X
2015	24	5	20.8	KA	0	0	1		
2014	65	9	13.8	L	1	0	1		
2013	39	3	7.7	M	0	0	0		
2012	38	4	10.5	N	0	0	1		
2011	106	12	11.3	O	2	2	5	1?	
2010	142	11	7.8	P				1?	
Total	**410**	**42**	**10.2**						

Present study – other studies

		2015 Simão	Caetano						2001 Simão	
Crom. Sexuais	**Azoo**									
	Oligo									
	Infer									
Autossomas	**Azoo**									
	Oligo									
	Infer									
Microdeleções Y	**Azoo**									
	Oligo									
	Infer									

Relatórios tenho

Nº	Data	Entidade	Indicação	Cariotipo	Estudos Y FISH	Result Y
	2010					
35639			Infertilidade	47,XYY		
60380	15.01	MACCAndro **38an**	Azoospermia secretóri→ICSI	46,XY		

66641		Tiago Rocha **41 anos**	Hipogo S Kline?	47,XXY		
68714	08.03	MAC ConAndro 34a	Para ICSI	46,XY		
70910	19.03	CSBenfica	Inf casal	46,XY,inv(1)(p13q12) Inv variante	**FISH SRY crom X**	
74242	09.04		S kline? 41 anos	46,X,der(X)t(X;Y)(p22.33;p11.3).ish der(X)t(X;Y)(p22.33;p11.3).ish der(X)t(X;Y)(p22.33;p11.3)(SRY+) (wcpY+;SRY+)	FISH	
74900	14.04	MAC **CapFerti 29an**	InfertilPrimária	46,XY	74901	
76981	27.04	MAC **CapFerti 41an**	Não indicado	46,XY	76982	
79446			AzoospSecretór	46,X,der(X)t(X;Y)(p22.33;p11.3).ish der(X)t(X;Y)(p22.33;p11.3)(wcpY+;SRY+)	FISH SRY no cromos X	
80750	19.05	MAC Candro **31an**	Hipogonadismo a esclarecer(S.Kline?)	**47,XXY**		
116498	27.12		OTA	47,XYY		
	2011					
118775		MAC C Androlo37	OTA	46,XY,t(4;14;15)	**FISH**	
118779		MACConApFerti 38 anos.Baixa estatur	Infertilidade	46,X,+mar.ish der(Y)(wcpT+,DYZ3+)	**FISH**	
122721	31.01	MACCApFert 42a	teratozoosperm	46,XY		
125376			Oligozoospermia	46,XY,rob(13;14),+mar.ish der(16)	**FISH Wcp16**	
126404	22.02	H Évora	OTA	46,XY		
127074	25.02	MAC CapFerti 31a	Azoospermia	46,XY		
127365	01.03	CS ArrudaVinhos	AR não entra	46,X,inv(Y)		
128576	10.03	H Évora	Azoospermia	46,XY		
127384	01.03	AVAClinic	Oligoteratozoosper	46,XY	127387	
128999	14.03	CSFaro- IVI	Infertilidade	46,XY	S	
129174		MAC CapFert 41a	Oligozoospia acentuada	46,XY	129175	
133654	13.04	HLusiadas CInfert	Azoospermia	46,X,inv(5),+mar[32]/45,X,inv(5[18]).ish i(Y)(p10)	133657 Tenho relatório **FISH**	AZFabc Ausente
136917			Azoospermia	mos 47,XXY[13]/46,XX[37]		
137344		15 anos não entra		47,XXY		
137431		Azoospermia		46,X,der(X)t(X;Y)(p22.31;p11.2)	FISH SRY no X	
138967			Azoospermia	46,XY,dup(8)(p23.1p23.1)	**FISH wcp8**	
142985	17.06	MAC **CApFert 29anos**	OTA	46,XY	142986	
146671			OTA	mos 47,XXY[2]/46,XY[48]		
156217		**32 anos**	Infertilidade primária Tia alter crom.	46,XY,t(7;22)(q11.23;q11.2)	156218	
157492	14.10	H Lusíadas	OTAacent→ICSI	46,XY	S	
159273	28.10	CS Parede	Teratozoospermi	46,XY	N	
164370			Azoospermia	mos 47,XXY[41]/46,XY[9]		
	2012					
175079	20.02	MAC C Andro **39 anos**	Azoospermia	46,XY	tem	
182396			Oligozoospermia	46,XY,t(4;8)(q27q11.23)		
203679			InfertPrimária	46,XX,inv(14)	**FISH**	
228218	14.12	HGOCirmaCApFer	não indicado	46,XY	228225	
	2013					
270209	05.07	HGOCirma	Nindicado?	47,XXY	?	
285260	01.10	HGOCirmaCApFert	Infertilidade	46,XY	?	
293890	11.11	HGOCirmaCap Fert	NãoInd/ Inferti	46,XY 36 anos	?	
297489	26.11	HGOCirmaCApFer	NãoInd/Inferti	46,XY 27 anos	297493	
300435	09.12	IVI	NI 7 Infert?	46,XY 38 anos	300436	
	2014					
340571	20.06	HGOCirmaCApFer 34 anos	Infertilidade	47,XY,+mar.ish der(14/22)D14Z1/D22Z1+)	**FISH**	

373887	26.11	HGO **CEx Infert**	Não ind	46,XY	373890	
376654	09.12	CHLC MAC Andro30an	**Infert Masc**	46,XY	376655	
380562	26.12	HGOCIRMA ApFert	46,xx Não			
	2015					
383373 **FOTO**	12.01	HGO CApFerti	Não indicado	46,XY,t(8;14) (p23.3;q11.2)	383374	
400626	10.03	HGOCirma **CApInf**40a	Sem ind		400629	

Abnormal Karyotypes

	2015 (até 30.03)				**Estu Y**	**Del AZFa**	**Del AZFb**	**Del AZFc**	**S R Y**
383373	**12.01**	**HGO CIRMA**	**infertilidade**	**46,XY,t(8;14)**	**383374**	-	-	-	

háFoto		CAFert		(p23.3;q11.2)					
386857	27.01	H Lusíadas	Infertilidade	46,XY,t(1;10)	386858	-	-	-	
399891	05.03	HDE	Infertilidade masculina	45,XY,rob(13;14)(q10;q10)	399894	-	-	-	
400626	10.03	HGO CIRMA	Infertilidade	46,XY	400629	-	-	deleção	
405321	26.03	ARS Alentejo	Infertilidade	46,XY,t(4;22)(p10;q10)	421323	-	-	-	
406016	30.03	HDE	Infertilidade	47,XXY	416018	-	-	-	
24casos				5 anomalias = 20,8%		0	0	1	
	2014								
307991	21.01	HEgasMoniz	Azoospermia	47,XXY	N				
340571	20.06	HGO CIRMA CExInfertili	infertilidade	47,XY,+mar.ish der(14/22)(D14Z1/D22Z1+)	340574	-	-	-	
343092	03.07	Hgo	infertilidade	46,XY,t(1;3)(q42.3;q26.2)	N				
345958	17.07	HGO	infertilidade	47,XXY	345959	-	-	-	
349769	01.08	Chlc	azoospermia	47,XXY	349770	-	-	-	
368774	04.11	HGO	azoospermia	Mos 45,X[34]/46,X,idic(Y)(q11.221)[16]	N				
373887	26.11	HGO	azoospermia	46,XY	373890	deleçã	-	-	
376654	09.12	CHLC mac	azoospermia	46,XY	376655	-	-	deleção	
379364	18.12	MAC CHLC c.And	Infertilidade pri	47,XXY	379366	-	-	-	
380104	22.12	CHLC	Infertilidade	46,XY,inv(8)???	380103	-	-	-	
380592	29.12	CHLC	infertilidade	47,XXY	380594	-	-	-	
2014		65 casos	9 anomalias	13,8% anomalias		1		1	
	2013	REVISTO nossos c/ Y							
255601			Susp S Klinefelter NÃO ENTRA - 17 anos	47,XXY não entra	N				
270209 X	05.07	HGO tb pede FQ	Infertilidade	47,XXY	270211	-	-	-	
271413	11.07	HDE Genética	Estudo pré-concecional Hist familiar. Pede drepanocitose	46,XY,inv(2)(p11.2q13) Não entra?	271414	Não i	o	vi	
293047	06.11	HGO	lifertilidade	47,XXY	293050	-	-	-	
		39 casos	3 anomalias =	7,7% anomalias		0	0	0	
	2012	Não fui ver outros "Não indicado" sem estudo Y	*Aqui estão todos de*	*Lista mercearia*	*B Molecular*				
175079	20.02	MAC	Azoospermia	46,XY	175080	-	-	deleção	
175993	24.02	CS ACES Oeste Norte	Infertilidade	47,XXY	175994	-	-	-	
182396 x	05.04	CHLC há FOTO	Infertilidad-OligoAcent	46,XY,t(4;8)(q27;q11.23)	182397	-	-	-	
188227	16.05	consultório	infertilidade	47,XXY	188228	-	-	-	
203679 x	14.08	H évora	Infertilidade primária	46,XY,inv(14)	N				
	46,XY,	Inv(14)(q13q22).ish	Inv(14)(p21.1)(RP11-388M7+)(q21.3)	(RP11-168D12+), *14q23.3(RP11-430G13x2)*					
Total	2012	41 casos (6 não entram)	4 Cariot anormal	9,8% anomalias	Nº Y =	0	0	1	
	2011								
164370 x	15.12	MAC	Azoospermia	mos 47,XXY[41]/46,XY[09]	N				

159998	**07.11**	**Particular - consultorio**	**Infertilidade azooospermia**	**47,XXY**	159999	-	-	-	
156217 x	**03.10**	**Arslvt lisboriental**	***Infertilidade primária*** *Tia com alter cromos.*	***46,XY,t(7;22)(q11.23;q11.2)***	156218	-	-	-	
146671 x	**14.07**	**ARSLVT**	**OTA oligoastenoteratozoos**	**mos 47,XXY[2]/46,XY[48]**	N				
142985	17.06	MAC	OTA	46,XY	142986			**gr/gr**	
138967 x	**19.05**	**MAC**	**Azoospermia**	**46,XY,dup(8)(p23.1p23.1). ish (wcp8+)**	138968	-	-	-	
137431 x	**10.05**	**HESanto Évora**	**Azoospermia**	**46,X,der(X) SRY está crom. X**	137434	**deleçã**	**deleçã**	**deleçã**	**No Cr. X**
		46,X,der(X)t(X;Y)(p22.31;p11.2).	ishder(X)t(X;Y)(p22.31;p11.2)(SRY+)						
137344 x	09.05	ARSLVTAcesLisboaNort	Desenv sex diminuido **NÃO ENTRA**	47,XXY	N				
136917 X	**05.05**	**MAC**	**Azoospermia**	**mos 47,XXY[13]/ 46,XX[37]**	136918	-	-	-	
133072	**08.04**	**MAC**	**Azoospermia**	**47,XXY**	133075	-	-	-	
133654	**13.04**	**ADSE**	**Azoospermia**	**46,X,+mar,inv(5)**	133657	**deleç**	**deleç**	**deleç**	
		46,X,+mar,inv(5)	(p14.2p15.2)[32]/ 45,X,inv(5)(p14.2 p15.2)[18]. Ish i(Y)	(p10)(SRY++,DYZ3+),inv(5)(wcp5+)					
130636	**24.03**	**MAC**	**Azoospermia**	**47,XXY**	130637	-	-	-	
129174 X	15.03	MAC	OligozoosperAcentu OTA	46,XY	129175	-	-	**deleção**	
127384	01.03	consultorio	infertilidade		127387	-	-	**deleção**	
125161	14.02	MAC	OTA	46,XY	125162	-	-	-	
125376	**15.02**	**MAC**	**Oligozoospermia**	**46,XY,rob(13;14)+mar**	N				
			46,XY,rob(13;14)+mar.	**Ish der(16)(wcp16+)**					
119484	13.01	MAC	criptozoospermia	46,XY	119486	-	-	**deleção**	
118775	**11.01**	**MAC**	**OTA**	**46,XY,t(4;14;15)**	N				
46,XY,	T(4;14;15)	(q22;q21;q21.39??).	Ish der(4)t(4;15)(wcp15+)	,der(14)t(4;14)(wcp4+),der(15)t(14;15)					
118779	**11.01**	**MAC**	***Infer, 38 anos baixa estatura***	***46,X,+mar.ish der(Y) (wcpY+,DYZ3+)***	N				
Em	2011	**106 /104 casos** (4 não entram)	**13 anomalias** (1 não entra)	**12,3% anomalias**	**Y = %**	**2**	**2**	**5**	
								gr/gr	
	2010								
60379	15.01	MAC	Azoosperm secretória	46,XY	60380	-	-	**deleção**	
62688	27.01	ARSLVT Oeste Sul	AR não entra 46,xy **Azoospermia**	46,XY	62689	-	**deleçã**	**deleção**	
65148	**11.02**	**ARSLVTACESOesteNort**	**Infertilidade**	**46,XY,rob(13;14)(q10;q10)**	N				
66641	23.02	TiagoR e CS St Isabel **Nãoentra? 42anos**	Hipogonadismo a esclarecer Suspeita Klinefelter?	*Mos 47;XXY[48]/46,XY[2]*	N				
68520	05.03	MAC	Azoospermia por	47,XXY	N				

			hipogonadismo primário						
70910	19.03	ARSLVT ACESLisboaNort	Esposa 2AE **Não entra**	46,XY,inv(1)(p13q12) variante	N				
74242 x	09.04		Suspeita S Klinefelter Não conta? 41 anos	*46,X,der(X)t(X;Y(p22.33;p11.3)*	N				
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33;;p11.3). ish der(X)t(X;Y)	(p22.33;p11.3)(SRY+)					
74900	14.04	MAC	infertilidade	46,xy	74901	-	**deleçã**	-	
76981		MAC	infertilidade	46,XY	76982	-	-	**deleção**	
79446	**10.05**	**MAC**	**Azoospermia secretória**	**der(X)t(X;Y(p22.33;p11.3) (wcp**	79447	**deleçã**	**deleçã**	**deleção**	**Ausente Y**
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33).ishder(X)t(X;Y)	(p22.33;p11.3)(wcpY+;SRY+)					
80750	**19.05**	**MAC**	**Hipogonadismo primário Susp S. Klinefelter ???**	**47,XXY**	N				
84664	**14.06**	**MAC**	**Criptozoospermia**	**47,XXY**	84668	-	-	-	
91268	**12.07**	**MAC**	**Azoospermia. Hipogonadismo primário**	**47,XXY**	91269	-	-	-	
95875	**12.08**	**MAC**	**Azoospermia**	**47,XXY**	N				
101373	**17.09**	**MAC**	**Azoospermi secretória**	**47,XXY**	101376	-	-	-	
116498	**27.12**	**MAC**	**OTA**	**47,XYY**	N				
Se não entrar com	**2015**	**Devido desvirtuar do HGO não indicados**	**Perco 4 translocações e XXY**						
383373	12.01	HGO	Não indicado	46,XY,t(8;14)	383374				
386857+	27.01	H Lusíadas	Infertilidade	46,XY,t(1;10)	386858				
399891+	05.03	HDE	Infertilidade	45,rob(13;14)(q10;q10)	399894				
405321+	26.03	ARS Alentejo	Infertilidade	46,XY,t(4;22)(p10;q10)	421323				
406016+	30.03	HDE	Infertilidade	47,XXY	416018				

Inférteis

Patologias		
AZO- Azoospermia		
OTA- Oligoteratoastenozoospermia		
OLI- Oligozoospermia		
OT- OligoTeratozoospermia		
TER- Teratozoospermia		
AO- Astenooligozoospermia??		

Deleções Y com % de Y em relação total casos e por indicações_____

178965 não nossa; 240506 não nossa, 318426 não nossa

	2015 (até 30.03)				**Casos c/ est Y Ndel (%)**	**Estu Y**	**Del AZFa**	**Del AZFb**	**Del AZFc**	**S R Y**
400626	10.03	HGO CIRMA	Infertilidade	46,XY		**400629**	-	-	Deleçã	

									7 posiç	
		23 casos		5 anomalias = 20,8%	15 1 (6,7)		0	0	1	
	2014									
373887	26.11	HGO	Azoospermia	46,XY		373890	Deleçã 6 posiçõ	-	-	
376654	09.12	CHLC mac	Azoospermia	46,XY		376655	-	-	A gr1	
	2014	64 casos	9 anomalias	13,8% anomalias	42 2 (4,8)		1		1	
	2013	38 casos	3 anomalias =	7,7% anomalias	23 0 (0)		0	0	0	
	2012									
175079	20.02	MAC	Azoospermia	46,XY		175080	-	-	Deleçã 4 posiçõ	
Total	2012	38 casos (6 não entram)	4 Cariot anormal	9,8% anomalias	31 1 (3,2)	Nº Y =	0	0	1	
	2011									
142985	17.06	MAC	OTA	46,XY		142986			gr/gr	
				Confirmar posi A gr		Em	AZFa	ou	AZFc	
137431	10.05	HESanto Évora	Azoospermia	46,X,der(X)t(X;Y) (p22.31;p11.2) SRY está crom. X		137434	deleçã	deleçã	deleçã	No Cr. X Vi
133654	13.04	Hosp Lusíadas- Adse	Azoospermia	46,X,inv(5),+mar[32]/ 45,X,inv(5)[18].ish i(Y)		133657	deleç	deleç	deleç	
129174	15.03	MAC	OligozoosperAcentu OTA	46,XY		129175	-	-	deleçã	
127384	01.03	consultorio	infertilidade	46,XY		127387	-	-	deleçã	
119484	13.01	MAC	criptozoospermia	46,XY		119486	-	-	deleçã	
Em	2011	106 casos (4 não entram)	13 anomalias (1 não entra)	12,3% anomalias	75 6 (8,0)	Y = %	2	2	5	
									gr/gr	
	2010									
60379	15.01	MAC	Azoospermia secretória	46,XY		60380	-	-	deleçã	
62688	27.01	ARSLVT Oeste Sul	AR não entra 46,xy Azoospermia	46,XY Não entra???		62689	-	deleçã	deleçã	
68666	08.03	ARSLVTAces Cacém-Qu	Não indicado Infertilidade	46,XY		68667	-	-	deleçã	
74900	14.04	MAC	infertilidade	46,XY		74901	-	deleçã	-	
76981	27.04	MAC C Ap ferti 41 anos	infertilidade	46,XY		76982	-	-	deleçã	

79446 X	10.05	MAC	Azoospermia
			secretória
				der(X)t(X;Y(p22.33;
				p11.3).ish
				der(X)t(X;Y)(p22.33;
				p11.3)(wcpY+;SRY+)
						79447	deleçã	deleçã	deleçã	No
										Cr
										X

	2010		? anomalias		71					

					6 (8,5)					
Totais		**410 casos**			**%**					

Estudos Y totais BM deles 2012

2012 casos	**SRY Pre sent**	**SRY Alte ração**	**Gr/gr c**	**AZFa**	**AZFb**	**AZFc**	**2012 Anormais total**				
110 (107)								*Totais Azo*	*12*		*delAZFb+c+a*	
AZO 13		x	xxx	x	x	xxx	9			*1*	*Del gr/gr*	
OLIGO 10			x			x	2			*1*	*Del AZFc*	
OTA 29			xxxx		x	xxxxx	10			*11*	*Normais (sem del AZF)*	
OT 3							0	Totais OLI	20		Del AZFc	
AO 5							0			2	Del gr/gr	
TERA 1							0				Normais (sem del AZF)	
Inf 45			x			xx	3					
Cont est 2			xx		xx	xx	6					
EMQN 3												
Total 108		1	11	1	4	13	30					

Casos com cariotipo anormal entre 2010 - Março 2015 (5 anos e 3 meses)

(Resumo) Não incluí AR, AE; Suspeita Klinefelter em jovens e adultos; Incluí casal; RMA;

Klinefelter; translocações; inversões; mosaicismos; marcadores;

Totais		**47,XXY** **47,XYY** **trans** **inv** **mar** **mos** **inv mar mos** **der (trans)**	**2** **12 rob:1** **5**	

ANOMALIAS Cromossómicas e Y no período em estudo

Estatística Sangues (+ tenho relatório)

	2015 (até 30.03)				Estu Y	Del AZFa	Del AZFb	Del AZFc	S R Y
381836		HGO CIRMA	Infert casal	46,XY	N				
382116	06.01	HGO CIRMA	Infertilidade	46,XY	382119	-	-	-	
382123	06.01	HGO	Azoospermia	46,XY	382124	-	-	-	
383373 háFoto	**12.01**	**HGO CIRMA CAFert**	**infertilidade**	**46,XY,t(8;14) (p23.3;q11.2)**	**383374**	-	-	-	
384417	15.01	CHLC	Infertilidade	46,XY	384419	-	-	-	
384933	19.01	H Lusíadas	Azoospermia	46,XY	N				
386857	**27.01**	**H Lusíadas**	**Infertilidade**	**46,XY,t(1;10)**	**386858**	-	-	-	
390424	30.01	CHLC - MAC	Azoospermia	46,XY	390425	-	-	-	
391101	03.02	HSJosé	Azoospermia. Hipogonadismo hipergonadotrófico	46,XY	N				
392675	10.02	HGO CIRMA	Infertilidade	46,XY	392677	-	-	-	
393432		HGO CIRMA	Infert casal	46,XY	N				
395324	19.02	HGO	Infertilidade	46,XY	395327	-	-	-	
396396		HGO CIRMA	Infert casal	46,XY	N				
399891	**05.03**	**HDE**	**Infertilidade masculine**	**45,XY,rob(13;14) (q10;q10)**	**399894**	-	-	-	
400497	09.03	HDE	Programa RMA casal	46,XY	N				
400626	10.03	HGO CIRMA	Infertilidade	46,XY	400629	-	-	deleção	
402080		ARSCentro Leiria	Não indicado	46,XY	N				
402701	17.03	HGO CIRMA	Infertilidade	46,XY	402705	-	-	-	
404339	23.02	AVAClinicaConsultorio	Infertilidade	46,XY	404345	-	-	-	
405321	**26.03**	**ARS Alentejo**	**Infertilidade**	**46,XY,t(4;22) (p10;q10)**	**421323**	-	-	-	
405784	27.03	HDE	Infertilidade primária	46,XY	405787	-	-	-	
406016	**30.03**	**HDE**	**Infertilidade**	**47,XXY**	**416018**	-	-	-	
406283		HGO CIRMA	Não indicado	46,XY	N				
406305		HGO CIRMA	Não indicado	46,XY	N				
23 24 casos			**24**	**5 anomalias = 20,8%**		**0**	**0**	**1**	
		Azoospermia **Infertilidade** **Infertilidade casal** **Não indicado**	**4** **13** **4** **3**	**0 anomalias** **5: 1 numé, 4 estru** **0** **0**				**0** **1** **0** **0**	
	2014								
305601	09.01	ARSN,IP Porto	Azoospermia	46,XY	305603	-	-	-	
307991	21.01	HEgasMoniz	**Azoospermia**	**47,XXY**	N				
310977	28.01	HGO CIRMA Infertili	Não indicado	46,XY	310978	-	-	-	
312606	04.02	CSMafra	Infer casal, ela 46,XX	46,xy	N				
318617	03.03	consultorio	Azoospermia	46,xy	318618	-	-	-	
319642	06.03	HGO CIRMA Infertili	Infertilidade	46,XY	319643	-	-	-	
322975	21.03	Chlc	Azoospermia	46,XY	N				
323537	25.03	proprio	Azoospermia	46,XY	323538	não	o	vi	
327021	10.04	ARS do Norte	Infertilidade	46,XY	327020	-	-	-	
329075	23.04	HGO	Infertilidade	46,XY	329076	-	-	-	
329633	28.04	Chlc	História infertilidade	46,XY	N				
330420	30.04	HGO	Infertilidade	46,XY	330421	-	-	-	
331145	05.05	CHLC	Azoospermia	46,XY	331146	-	-	-	
331579	06.05	CHLC	Infertilidade	46,XY	331580	não	o	vi	
332641	12.05	HGO	Não indicado	46,XY	332642	não	o	vi	
333437	14.05	HGO	Infertilidade	46,XY	333440	-	-	-	
336073	27.05	CHLC	Infertilidade casal	46,XY	N				
336112	28.05	CHLC	Infertilidade	46,XY	N				
336153	28.05	Proprio - consultorio	Infertilidade	46,XY	N				
337192	02.06	CHLC	Infert hipogonadismo	46,XY	337193	-	-	-	
339619	17.06	HGO	Infert casal	46,XY	N				

340180	19.06	HGO Cirma ApInfert	Infertilidade	46,XY	340185	-	-	-	
340571	20.06	HGO CIRMA CExInfertili	infertilidade	47,XY,+mar.ish der(14/22) (D14Z1/D22Z1+)	340574	-	-	-	
341443	26.06	CHLC	OTA	46,XY	341445	-	-	-	
341961	30.06	proprio	Infertilidade	46,XY	N				
342326	01.07	proprio	Infer casal	46,XY	N				
342400	01.07	Chlc- mac	Infertilidade	46,XY	342401	-	-	-	
342952	02.07	Chlc	Infertilidade casal	46,XY	N				
343092	03.07	Hgo	Infertilidade	46,XY,t(1;3)(q42.3;q26.2)	N				
344882	11.07	Chlc	Azoospermia	46,XY	344884	-	-	-	
345958	17.07	HGO	**Infertilidade**	**47,XXY**	345959	-	-	-	
349769	01.08	Chlc	**Azoospermia**	**47,XXY**	349770	-	-	-	
350999	07.08	CHLC mac	Infert hipogonadismo	46,XY	351001	-	-	-	
351456	11.08	Chlc	Azoospermia	46,XY	351459	-	-	-	
351543	12.08	Subsistem consultorio	Infertilidade	46,xy	351544	-	-	-	
352380	18.08	Consultorio	Infertilidade	46,XY	352382	-	-	-	
353906	26.08	Hgo	Infert	46,xy	N				
354462	28.08	**Hgo**	Infertilidade	46,xy	N				
354637	29.08	**HGO**	In casal	46,XY	N				
356580	09.09	**HGO**	Infertilidade	46,XY	356582	-	-	-	
356829	10.09	ARSLVT	Azoospermia	46,XY	356831	-	-	-	
357158	11.09	CHLC	Infert hipogona	46,XY	357164	-	-	-	
357485	12.09	Subsistemas H Luz	Infer casal	46,XY	N				
357537	12.09	HGO	Azoospermia	46,XY	357538	-	-	-	
358746	18.09	CHLC mac	Infertilidade hipogon	46,XY	358747	-	-	-	
360154	23.09	CHLC	Azoospermia	46,XY	N				
360614	25.09	H Lusiadas	Não indicado	46,XY	360611	não	o	vi	
363876	10.10	CHLC	Azoospermia	46,XY	363877	-	-	-	
365605		HGO	Infertilidade	46,XY	365606	-	-	-	
365952	22.10	HGO	Infertilidade	46,XY	365953	-	-	-	
366308	23.10	CHLC	Infert hipogo	46,XY	366309	-	-	-	
368774	04.11	HGO	Azoospermia	**Mos 45,X[34]/46,X,idic(Y)(q11.221)[16]**	N				
369793	07.11	consultorio	Infer casal	46,XY	369795	não	o	vi	
370952	12.11	CHLC	Infertilidade primária – casal	46,XY	N				
371273	13.11	HGO	Não indicado	46,XY	371310	não	o	vi	
372797	20.11	CHLC mac	Infertilidade	46,XY	373061	-	-	-	
372860	20.11	CHLC	Infertilidade	46,XY	N				
373887	26.11	HGO	Azoospermia	46,XY	373890	deleçã	-	-	
374208	27.11	HGO	Infertilidade	46,XY	374213	-	-	-	
376643	09.12	CHLC	Infertilidade	46,XY	376644	-	-	-	
376654	09.12	CHLC mac	Azoospermia	46,XY	376655	-	-	deleção	
379364	**18.12**	**MAC CHLC c.And**	**Infertilidade pri**	**47,XXY**	379366	-	-	-	
379212	18.12	ARSLVT CSSeixal	Infertilidade casal	46,XY	379192	-	-	-	
380104	**22.12**	**CHLC**	**Infertilidade**	**46,XY,inv(8)???**	380103	-	-	-	
380592	**29.12**	**CHLC**	**infertilidade**	**47,XXY**	380594	-	-	-	
		64 65 casos	**9 anomalias**	**13,8%anomalias**		**1**		**1**	
		Azoospermia **OTA** **Infertilidade** **Infertilidade casal** **Não indicado**	**16** 1 34 10 4	**3: 2 numé, 1 estMos** **6: 4 numé, 2 estru**		**1**		**1**	
	2013	REVISTO nossos c/ Y							
236412	23.01	CHMT	I casal?	46,XY	N				
237041	25.01	ARSLVT	I casal	46,XY	N				
240263	11.02	ULSMatosinhos	I casal	46,XY	N				
240942	14.02	CHSetúbal	OTA	46,XY	240943	-	-	-	

243155	26.02	ARSLVT-ACES Cascais	teratozoospermia	46,XY	N				
248520	20.03	ARSLVT-ACES Lisboa Norte	Azoospermia	46,XY	248521	-	-	-	
248968	21.03	Subsistemas - IVI	Infertilidade	46,XY	248970	-	-	-	
250209		HGO CIRMA CAInferti	Infertilidade	46,XY	250214	-	-	-	
255601			Susp S Klinefelter **NÃO ENTRA - 17 anos**	**47,XXY não entra**	N				
259097	07.05	CHLC	I casal	46,XY	N				
260632	15.05	HGO	Infertilidade	46,XY	260624	-	-	-	
261042	16.05	HGO	Infertilidade	46,XY	261043	-	-	-	
261777	21.05		Hipogonadismo hipogonadotrofico	46,XY	261779	não	o	vi	
261862	21.05	HGO	Infertilidade	46,XY	261865	-	-	-	
262024	22.05	ARSLVTACESAmadora	I casal	46,XY	N				
262059	22.05	HGO	Infertilidade	46,XY	262061	-	-	-	
263057	28.05	HGO	I casal	46,XY	N				
263236	29.05	Subsistemas	OTA	46,XY	263239	-	-	-	
265019	05.06	CHLC	I casal?	46,XY	N				
265525	07.06	HGO	I casal?	46,XY	N				
266057	12.06	HGO	I casal	46,XY	N				
267042	19.06	ULSBxA	I casal	46,XY	N				
267704	24.06	ARS Faro	I casal	46,XY	267705	-	-	-	
270209 X	05.07	HGO tb pede FQ	Infertilidade	**47,XXY**	270211	-	-	-	
270750	09.07	HGO CIRMA	Infertilidade	46,XY	270752	-	-	-	
270936	10.07	ARSLVT	I casal	46,XY	N				
271413	11.07	HDE Genética	Estudo pré-concecional Hist familiar. Pede drepanocitoser	**46,XY,inv(2)(p11.2q13) entra**	271414	Não i	o	vi	
273127	18.07	HGO CAInferti	Infertilidade	46,XY	273128	-	-	-	
282521	17.09	HGO CIRMA	Infertilidade	46,XY	282523	-	-	-	
283227	19.09	HGO	Oligozoospermia grav	46,XY	283228	-	-	-	
285260 X	01.10	HGO Cirma	Infertilidade	46,XY	285264	-	-	-	
292072	01.11	Subsistemas	Infertilidade	46,XY	292075	-	-	-	
292708	05.11	HGO	Infertilidade	46,XY	292710	-	-	-	
293024	06.11	HGO	Infertilidade	46,XY	293028	-	-	-	
293047	06.11	HGO	Infertilidade	**47,XXY**	293050	-	-	-	
293890 X	**11,11**	**HGO cario,micro,FQ CIRMA CAInfert**	**Infertilidade**	**46,XY**	**293893**	**-**	**-**	**-**	
297489 X	26.11	HGO cario.mic,FQ CIRMA CAInfert	Infertilidade	46,XY	**297493**	**-**	**-**	**-**	
300435 X	09.12	IVI	Infertilidade	46,XY	**300436**	**-**	**-**	**-**	
301906	16.12	ARSLVT	Infert casal	46,XY	N				
		38 casos	**3 anomalias**	**7,9 % anomalias**		**0**	**0**	**0**	
		Azoospermia Oligozoospermia OTA teratozoospermia Infertilidade Infertilidade casal Não indicado	1 1 2 1 19 14 0	**3: 2 numé, 1 estru**					
	2012	Não fui ver outros "Não indicado" sem estudo Y	***Aqui estão todos de***	***Lista mercearia***	***B Molecular***				
169458	13.01	MAC Cons. A Fertili	Oligozoospermia	46,XY	169459	-	-	-	
171114	26.01	CS- ACES Loures	Azoospermia	46,XY	171115	-	-	-	
172039	01.02	MAC	Oligozoospermia	46,XY	172040	-	-	-	
172531	03.02	MAC	Azoospermia	46,XY	172532	-	-	-	
172796	06.02	MAC	Oligozoospermia	46,XY	172797	-	-	-	
172965	07.02	MAC Cons Andro	Azoospermia	46,XY	172966	-	-	-	
173439	09.02	MAC	Oligoteratozoosper	46,XY	173440	-	-	-	
173939	13.02	MAC	OTA	46,XY	173941	-	-	-	
174336	15.02	MAC	OTA	46,XY	174337	-	-	-	
174584	16.02	MAC	OTA	46,XY	174586	-	-	-	
175079	20.02	MAC	Azoospermia	46,XY	175080	-	-	deleção	

175163	20.02	MAC	OTA	46,XY	175164	-	-	-	
175756	23.02	MAC	OTA	46,XY	175757	-	-	-	
175993	**24.02**	**CS ACES Oeste Norte**	**Infertilidade**	**47,XXY**	175994	-	-	-	
177206	05.03	Subsistemas –particul	Infertilidade	46,XY	177207	-	-	-	
177401	06.03	CHLC	OTA	46,XY	177402	-	-	-	
177413	06.03	CHLC	OTA	46,XY	N				
181661	03.04	CHLC	OTA	46,XY	181662	-	-	-	
181924	04.04	consultório	infertilidade	46,XY	181926	-	-	-	
182396 x	**05.04**	**CHLC há FOTO**	**Oligozoospermi**	**46,XY,t(4;8)(q27; q11.23)**	182397	-	-	-	
182436	09.04	CHLC	OTA	46,XY	182437	-	-	-	
183648	16.04	CHLC- MAC	OTA	46,XY	183649	-	-	-	
185639	30.04	ARSLVTLisb?CSSomagu	infertilidade	46,XY	185640	-	-	-	
186413	04.05	CHLC- MAC	Azoospermia	46,XY	186414	-	-	-	
186579	07.05	consultório	AR t(10;22) **não entra** Infertili???		186580	-	-	-	
188227	16.05	consultório	**infertilidade**	**47,XXY**	188228	-	-	-	
193389	14.06	ARSLVAcesOeiraUSFdel t	Infertilidade	46,XY	N				
199869	23.07	CHLC	AE não conta 46,xy		199870	-	-	-	
200009	24.07	Hévora	Infertilidade primária	46,XY	N				
201199	31.07	Hévora	Infertilidade primária	46,XY	N				
203679 x	**14.08**	**Hévora**	**Infertilidade primária**	**46,XY,inv(14)**	N				
	46,XY,	**Inv(14)(q13q22).ish**	**Inv(14)(p21.1)(RP 11-388M7+)(q21.3)**	**(RP11-168D12+), *14q23.3(RP11-430G13x2)***					
208214	04.09	ARSLVTAces0eir/HCCabr	Infertilidade	46,XY	208213	-	-	-	
208779	06.09	CHLO HegasMoniz	AR 46,xy não entra		208779	não	o	vi	
213686	04.10	CHLO	AE não entra 46,XY		213687	não	o	vi	
215392	15.10	hegas	casalAR não entra 46,xy		215393	não	o	vi	
216937	23.10	HGO CIRMA apoio inf	Infertilidade	46,XY	216938	-	-	-	
217491	25.10	HGO CIRMA apoioInfer	Infertilidade	46,XY	N				
220885	12.11		casalAR não entra46,XY		220886	não	o	vi	
221612	15.11	CHLO hde genética	3 perdas fetais Não entra 46,XY		221609	não	o	vi	
224983	30.11	British hospital	Alter espermograma	46,xy	224963	não	o	vi	
225194	03.12	ClinicaCUFTorresVedra	Azoospermia	46,XY	N				
225867	05.12	Hgo cirma	Infert casal	46,xy	N				
226083	05.12	CHLO HEgas	3 perda fetais não entra 46,xy Trombose		226084	não	o	vi	
227302	12.12	HGO cirma	Infertilidade	46,XY	227303	-	-	-	
228218	14.12	HGO cirma	Infertilidade	46,XY	228225	-	-	-	
228506	17.12	Subsiste HSantiagSetu	Infertilidade	46,XY	228508	-	-	-	
Total	**2012**	**38 casos** (8 não entram)	**4 Cariot anormal**	**9,8% anomalias**	**Nº Y =**	**0**	**0**	**1**	
		Azoospermia Oligozoospermia OTA teratozoospermia Infertilidade Infertilidade casal Não indicado	6 5 10 0 16 1 0	**1: 1 estru** **3: 2 numé, 1 estru**				**1**	
	2011								
165988	21.12	MAC	Azoospermia	46,XY	165989	-	-	-	
165496	19.12	MAC	Azoospermia	46,XY	165499	-	-	-	
164370	**15.12**	**MAC**	**Azoospermia**	**mos 47,XXY[41]/**	N				

x				**46,XY[09]**					
162753	05.12	MAC	Azoospermia	46,XY	162754	não	o	vi	
163015	06.12	MAC	Oligoastenozoosper AO	46,XY	163016	-	-	-	
161837	22.11	MAC CAFertili	Infertilidade FIV	46,XY	161838	-	-	-	
157492 x	14.10	ADSE consultorio	OTA acentd. para ICSI	**46,XY**	157506	-	-	-	
159273 x	28.10	ARSLVT- Oeiras	Teratozoospermia	46,XY	N				
159361	28.10	mac	Infertilidade primária	46,XY	159362	-	-	-	
159683	03.11	Particular	Infertilidade	46,XY	N	-	-	-	
159888	04.11	csOlivaisarslisbOriental	Infertilidade	46,XY	158889	-	-	-	
159998	**07.11**	**Particular - consultorio**	**Azooospermia**	**47,XXY**	159999	-	-	-	
160134	08.11	CSOeiras	Azoospermia	46,xy	160136	-	-	-	
160725	14.11	MAC	Azoospermia	46,XY	160726	-	-	-	
154398	21.09	MAC	oligospermia	46,XY	154399	-	-	-	
154855	22.09	MAC	OTA	46,XY	154856	-	-	-	
155637	28.09	ARSLVT	Azoosperm secretória	46,XY	155638	-	-	-	
156217 x	**03.10**	**Arslvt lisboriental**	***Infertilidade primária*** *Tia com alter cromos.*	***46,XY,t(7;22)(q11.23;q11.2)***	156218	-	-	-	
156378	04.10	MAC	Oligospermia	46,XY	156379	-	-	-	
156533	06.10	MAC	Oligospermia	46,XY	156534	-	-	-	
156703	07.10	MAC	azoospermia	46,XY	156704	-	-	-	
157021	11.10	MAC	OTA	46,XY	157023	-	-	-	
151107	24.08	Arslvt santarem	Infertilidade	46,xy	151108	-	-	-	
151225	25.08	Particular	Infertilidade casal	46,XY	N				
151593	30.08	MAC	OTA	46,XY	151594	-	-	-	
151910	01.09	MAC	OT	46,XY	151914	-	-	-	
152769	08.09	MAC Cons Andro 1976	Infertilidade	46,XY	152770	-	-	-	
152947	09.09	Mac ConsulAndrol1983	Infertilidade	46,XY	152948	-	-	-	
152963	09.09	mac	Infertilidade	46,XY	152965	-	-	-	
148131	26.07	ARSLVT-VFXira	Azoosperm secretória	46,XY	148133	-	-	-	
148857	01.08	MAC	OTA	46,XY	148858	-	-	-	
149608	08.08	MAC	Oligospermia (OT)?	46,XY	149609	-	-	-	
149879	10.08	MAC	Infertilidade mascu	46,XY	N				
150501	17.08	MAC	oligozoospermia	46,XY	150503	-	-	-	
145146	05.07	CHLN	Infertilidade	46,XY	N				
146096	11.07	MAC CAF	Infertilidade primá	46,xy	146099	-	-	-	
146441	13.07	MAC	Azoospermia	46,XY	146442	-	-	-	
146671 x	**14.07**	**ARSLVT**	**OTA Oligoastenoteratozoospermia**	**mos 47,XXY[2]/46,XY[48]**	N				
146910	18.07	MAC	Teratozoospermia	46,XY	146911	-	-	-	
146921	18.07	Arslvt santarem	Infertilidade	46,xy	146922	-	-	-	
146938	18.07	ARSLVTOesteNorte	Infertilidade	46,XY	146939	-	-	-	
147057	19.07	ARSLVT-Santarém	Infertilidade casal	46,XY	N				
144145	28.06	MAC	OTA	46,XY	144148	-	-	-	
142985	17.06	MAC	OTA	46,XY	142986			gr/gr	
144503	30.06	USF And	Infertilidade	46,XY	144504				
144354	29.06	British, USF AlphaMouro	Infertilidade	46,xi	144356	-	-	-	
140736	01.06	MAC	Azoospermia	46,XY	140737	-	-	-	
140778	01.06	MAC	Infertilidade	46,XY	140780	-	-	-	
140072	26.05	ARSLVT aces oeiras	Infertilidade	46,XY	140073	-	-	-	
140380	30.05	ARS Setubal	Azoospermia	46,XY	140381	-	-	-	
140383	30.05	MAC CAF	Infertilidade	46,XY	140384	-	-	-	
141429	06.06	arslvtejo	Inferti + rins poliquist	46,XY	131430	não	o	vi	
138258	16.05	ARS-ACESAlgarve	OTA	46,XY	N				
138597	17.05	MAC	azoospermia	46,XY	138598	-	-	-	
138967 x	**19.05**	**MAC**	**Azoospermia**	**46,XY,dup(8)(p23.1p23.1). ish (wcp8+)**	138968	-	-	-	
139483	23.05	MAC	Azoospermia	46,XY	N				

137235	09.05	MAC	Oligo acentu.Terato OTZ	46,XY	137236	-	-	-	
137270	09.05	MAC	OTA	46,XY	137271	-	-	-	
137431 x	**10.05**	**HESanto Évora**	**Azoospermia**	**46,X,der(X) SRY está crom. X**	137434	deleçã	deleçã	deleçã	No Cr. X
		46,X,der(X)t(X;Y)(p22.31;p11.2).	ishder(X)t(X;Y)(p22.31;p11.2)(SRY+)						
137631	11.05	MAC	OTA	46,XY	137632	-	-	-	
137344 x	09.05	ARSLVTAcesLisboaNort	Desenv sex diminuido **NÃO ENTRA 16 anos**	47,XXY	N				
137814	12.05	MAC	Teratozoospermia	46,XY	N				
138099	13.05	MAC	OTA	46,XY	138101	-	-	-	
134867	20.04	MAC	Azoospermia	46,XY	134868	-	-	-	
136917 X	**05.05**	**MAC**	**Azoospermia**	**mos 47,XXY[13]/ 46,XX[37]**	136918	-	-	-	
136417	03.05	MAC	Oligozoospermia	46,XY	N				
133072	**08.04**	**MAC**	**Azoospermia**	**47,XXY**	133075	-	-	-	
133421	12.04	MAC	oligozoospermia	46,XY	133422	-	-	-	
133654	**13.04**	**ADSE**	**Azoospermia**	**46,X,+mar,inv(5)**	133657	deleç	deleç	deleç	
		46,X,+mar,inv(5)	(p14.2p15.2)[32]/ 45,X,inv(5)(p14.2 p15.2)[18]. Ish i(Y)	(p10)(SRY++,DYZ3+),inv(5)(wcp5+)					
130636	**24.03**	**MAC**	**Azoospermia**	**47,XXY**	130637	-	-	-	
130789		HEgas	AE não entra46,XY		130792	não	o	vi	
131241	29.03	HESanto Évora	Infertilidade	46,XY	N				
131312	29.03	MAC	Criptozoospermia	46,XY	131313	-	-	-	
131989	01.04	MAC	teratozoospermia	46,XY	N				
132009	01.04	MAC	teratozoospermia	46,XY	132011	-	-	-	
128576 X	10.03	HESanto Évora	Azoospermia	46,XY	N				
128843	11.03	MAC	Azoospermia	46,XY	128844	-	-	-	
128999 X	14.03	ARSAlgarveAcesAlgarve	Infertilidade casal	**46,XY**	129000	Não	o	vi	
129174 X	15.03	MAC	OligozoosperAcentu OTA	46,XY	129175	-	-	deleção	
127924	03.03	HESanto Évora	OTA	46,XY	N				
128550	10.03	MAC	Astenoteratozoosper	46,XY	N				
126404 X	22.02	HESanto Évora	OTA	46,XY	N				
126466	22.02	ARSAcesLVT Norte CS Sete Rios	OTA	46,XY	126467	-	-	-	
126434	22.02	MAC	OTA	46,XY	126436	-	-	-	
127026	24.02	Hegas CHLO	AB e não entra 46,XY		127027	não	o	vi	
127074 X	25.02	MAC	Azoospermia	46,XY	N				
127384	01.03	consultorio	Infertilidade		127387	-	-	deleção	
125161	14.02	MAC	OTA	46,XY	125162	-	-	-	
125376 X	**15.02**	**MAC**	**Oligozoospermia**	**46,XY,rob(13;14)+mar**	N				
			46,XY,rob(13;14)+mar.	**Ish der(16)(wcp16+)**					
122721 X	31.01	MAC	TeratozoospermiaSev	**46,XY**	N				
123407	03.02	MAC	Infertilidade	46,XY	N				
124377	09.02	MAC	OTA	46,XY	124379	-	-	-	
124752			AE não entra		124754	Não i	o	vi	
121173	24.01	MAC	OTA	46,XY	121177	-	-	-	
121235 X	24.01	MAC CAF	OTA	46,XY	121238	-	-	-	
121622 X	25.01	MAC	Infertilidade	46,XY	121623	-	-	-	
119476	13.01	MAC	OTA	46,XY	119477	-	-	-	
119484	13.01	MAC	criptozoospermia	46,XY	119486	-	-	deleção	
119500	13.01	MAC	Azoospermia	46,XY	119501	-	-	-	
119627	14.01	MAC	OTA	46,XY	N				
119835	17.01	MAC	criptozoospermia	46,XY	119836	-	-	-	
120076	18.01	MAC	criptozoospermia	46,XY	120077	-	-	-	
120236	19.01	MAC	Infertilidade	46,XY	120237	-	-	-	
121021	21.01	ARSLVT AcesRibatejo	Infert casal	46,XY 46,XX	121022	-	-	-	
117417	03.01	MAC	OTA	46,XY	N				

117621	04.01	MAC	OTA	46,XY	117622	-	-	-	
117642	04.01	MAC	OTA	46,XY	117643	-	-	-	
118587	11.01	MAC	OTA	46,XY	N				
118775 X	**11.01**	**MAC**	**OTA**	**46,XY,t(4;14;15)**	N				
46,XY,	T(4;14; 15)	(q22;q21;q21.39??).	Ish der(4)t(4;15)(wcp15 +)	,der(14)t(4;14)(wcp4+), der(15)t(14;15)					
118779 X	**11.01**	**MAC**	***Infer, 38 anos*** *baixa estatura*	***46,X,+mar.ish der(Y) (wcpY+,DYZ3+)***	N				
Em	2011	**106 /104 casos** (4 não entram)	**13 anomalias** (1 não entra)	**12,3% anomalias**	**Y = %**	**2**	**2**	**5**	**x**
		Azoospermia Oligozoospermia OTA OT Astenoteratozoospe Teratozoospermia Criptozoospermia Infertilidade Infertilidade casal Não indicado	25 9 28 1 1 6					**1**	
	2010								
59150	08.01	MAC	Infertilidade	46,XY	N				
59239	08.01	CS Marvila	Azoospermia	46,XY	59240	-	-	-	
59828	13.01	MAC	Azoospermia	46,XY	59829	-			
60379	15.01	MAC	Azoosperm secretória	46,XY	60380	-	-	**deleção**	
60432	15.01	MAC	OTA	46,XY	N				
61120	19.01	ARSLVT CSSacavém	infertilidade	46,XY	61121	-	-	-	
61386	20.01	ARSLVT ACES Cacém	Teratozoospermia	46,XY	61387	Não	o	vi	
61658	21.01	ARSLVT ACES Amadora	Azoospermia	46,XY	61660	-	-	-	
62242	25.01	MAC	OTA	46,XY	N				
62442	26.01	MAC	Astenoteratozoosper	46,XY	N				
62446	26.01	MAC	Azoospermia secretór	46,XY	62447	-	-	-	
62688	27.01	ARSLVT Oeste Sul	AR não entra 46,xy **Azoospermia**	46,XY	62689	-	deleçã	**deleção**	
63070	28.01	HEgas	AE não entra 46,xy		63072	não	o	vi	
63149	29.01	MAC	ICSI	46;XY	N				
63173	29.01	MAC	Astenoteratozoosper	46,XY	N				
64036	04.02	MAC	Azoosperm secretória	46,XY	64037	-	-	-	
64717	09.02	MAC	OTA, ICSI	46,XY	N				
65148	**11.02**	**ARSLVTACESOesteNort**	**Infertilidade**	**46,XY,rob(13;14) (q10;q10)**	**N**				
65403		MAC	OTA	46,XY	N				
65992	18.02	HEgas	AE não entra 46,xy		65993	não	o	vi	
66234	19.02	MAC	Infertilidade	46,XY	N				
66641 X	**23.02**	**TiagoR e CS St Isabel**	***Hipogonadismo a esclarecer Susp Klinefelter? Nãoentra? 42anos***	***Mos 47;XXY[48]/46,XY[2]***	**N**				
68666	08.03	ARSLVTAces Cacém-Qu	Infertilidade	46,XY	68667			**deleção**	
66801	24.02	MAC	astenoteratozoosper	46,XY	N				
66808	24.02	ARSLVTCSSeixal	infertilidade	46,XY	66809	-	-	-	
66830	24.02	MAC	Azoospermia	46,XY	66831	-	-	-	
66070	25.02	ARSLVTACESLisboaNorte	Azoospermia obstrutiva 46,XY		67071	Não	o	vi	
67298	26.02	MAC	astenoteratozoospermia (para ICSI)	46,XY	N				
68520	**05.03**	**MAC**	**Azoospermia por hipogonadismo primário**	**47,XXY**	**N**				

68548	05.03	MAC	OTA	46,XY	68551	-	-	-	
68666	08.03	arsAcesLVTCacém-Qu	Infertilidade	46,XY	68667				
68714 X	08.03	MAC	ICSI	46,XY	N				
69117	09.03	MAC	Azoospermia - ICSI	46,XY	69118	-	-	-	
69528	11.03	MAC	astenoteratozoosper	46,XY	N				
69777	12.03	MAC	(astenoteratozoospermia) para ICSI	46,XY	N				
69882	15.03	MAC	Azoospermia obstrutiva *46,xy*		69883	-	-	-	
69936	15.03	MAC	astenoteratozoosper	46,XY	N				
70910 X	19.03	ARSLVT ACESLisboaNort	Esposa 2AE **Não entra**	46,XY,inv(1)(p13q12) variante	N				
71047	22.03	MAC	OTA	46,XY	N				
71226	23.03	MAC	Infertilidade	46,XY	N				
71526	24.03	MAC	OTA	46,XY	71528	-	-	-	
72413	29.03	ARSLVTCS Graça	infertilidade	46,XY	72414	-	-	-	
72897	31.03	MAC	OTA	46,XY	N				
74074			AR 46,XY		74077	não	o	vi	
74242 x	09.04		Suspeita S Klinefelter Não conta? 41 anos	***46,X,der(X)t(X;Y(p22.33;p11.3)***	N				
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33;;p11.3). ish der(X)t(X;Y)	(p22.33;p11.3)(SRY+)					
74322	09.04	ARSLVT	infertilidade	46,XY	74368	-	-	-	
74900	14.04	MAC	infertilidade	46,xy	74901	-	deleçã	-	
75161	14.04		Ar não conta46,xy		75162	não	o	vi	
75715	19.04	MAC	OTA	46,XY	N				
75735	19.04	MAC	infertilidade	46,XY	75739	-	-	-	
76180	21.04	MAC	OTA para ICSI	46,XY	N				
76615	23.04	CSAbrantes	infertilidade	46,XY	76624	-	-	-	
76638	23.04	MAC	Astenoteratozoosper	46,XY	N				
76981		MAC	infertilidade	46,XY	76982	-	-	deleção	
77116	27.04	MAC	infertilidade	46,XY	77117	-	-	-	
77495	29.04	MAC	OTA c/ indicação para ICSI	46,XY	N				
78041	03.05	MAC	OTA	46,XY	N				
78263	03.05	MAC	OTA	46,XY	78264	-	-	-	
79446 X	**10.05**	**MAC**	**Azoospermia secretória**	**der(X)t(X;Y(p22.33;p11.3) (wcp**	79447	deleçã	deleçã	deleção	Ausente Y
		46,X,der(X)t(X;Y(p22.33;p11.3).ish der(X)t(X;Y)	(p22.33).ishder(X)t(X;Y)	(p22.33;p11.3)(wcpY+;SRY+)					
79741	12.05	MAC	OTA	46,XY	N				
80750	*19.05*	*MAC*	*Hipogonadismo primário SuspS.Klinefelter?*	*47,XXY*	*N*				
80763	19.05	MAC	Astenoteratozoosper	46,XY	N				
81777	25.05	MAC	Azoospermia	46,XY	81778	-	-	-	
82138	26.05	MAC	OTA	46,XY	N				
82716	28.05	MAC	ICSI	46,XY	N				
83621	04.06	MAC	Azoospermi secretória	46,XY	83622	-	-	-	
83633	04.06	MAC	Criptozoospermia	46,XY	83634	-	-	-	
83744	07.06	MAC	OTA	46,XY	83745	-	-	-	
84315	09.06	ARSLVT	Azoospermia	46,XY	84316	-	-	-	
84517	11.06	ARSLVTACESLisboaCen	Azoospermia	46,XY	84518	-	-	-	
84664	**14.06**	**MAC**	**Criptozoosperm**	**47,XXY**	84668	-	-	-	
84750	14.06	MAC	Azoospermia	46,XY	84751	-	-	-	
85905	17.06	MAC	OTA	46,XY	N				
86169	17.06	MAC	OTA	46,XY	86170	-	-	-	
86836	22.06	MAC	OTA	46,XY	86837	-	-	-	
86871	22.06	MAC cons. FIV	infertilidade	46,xy	86872	-	-	-	
87953	25.06	ARSLVT-ACES Setúbal	Oligoteratozoosperm	46,XY	N				
88098	28.06	MAC	Criptozoospermia	46,XY	88099	-	-	-	
88383	29.06	MAC	teratozoospermia	46,XY	N				

88393	29.06	MAC	Azoospermia	46,XY	88394	-	-	-	
90203	06.07	ADSE	Oligoteratozoospermi	46,XY	N				
91246	12.07	MAC	teratozoospermia	46,XY	N				
91268	**12.07**	**MAC**	**Azoospermia. Hipogonadismo primário**	**47,XXY**	91269	-	-	-	
91653	13.07	ARSLVT	infertilidade	46,XY	91654	-	-	-	
92281	15.07	MAC	Teratozoospermia	46,XY	N				
92601	19.07	MAC	Astenoteratozoosper	46,XY	N				
92719	20.07	ARSLVT ACES Leziria I	Azoospermia	46,XY	92720	-	-	-	
93502	26.07	MAC	OTA	46,XY	93503	-	-	-	
94111	29.07	MAC	Criptozoospermia	46,XY	94112	-	-	-	
94464	02.08	Consultório	OTA	46,XY	N				
94764	04.08	MAC	ICSI	46,XY	N				
95127	06.08	MAC	OTA	46,XY	95128	-	-	-	
95875	**12.08**	**MAC**	**Azoospermia**	**47,XXY**	N				
96357	16.06	MAC	Azoosperm secretória	46,XY	96359	-	-		
96867	19.08	MAC	Asteno e Terato severas	46,XY	96868	Não	o	vi	
97603	24.08	MAC	OTA	46,XY	N				
98122	27.08	MAC	Azoospermia	46,XY	98123	-	-	-	
98192	27.08	MAC	Teratozoospermia c/ discreta astenozoosp	46,XY	N				
98483	30.08	MAC	OTA	46,XY	N				
98485	30.08	MAC	Teratozoospermia	46,XY	N				
99156	02.09	ARSLVTCSMontijo	infertilidade	46,XY	99157	-	-	-	
99199	02.09	MAC	Azoospermia	46,XY	N				
100954	15.09	MAC	OTA	46,XY	N				
101373	**17.09**	**MAC**	**Azoospermi secretória**	**47,XXY**	101376	-	-	-	
101459	17.09	MAC	OTA	46,XY	N				
101610	20.09	MAC	Astenoteratozoosper	46,XY	N				
101642	20.09	MAC	Infertilidade	46,XY	N				
101926	21.09	CHLO	Susp S Klinefelter **não entra– 17 anos**	46,XY	N				
102329	23.09	MAC	OTA	46,XY	N				
102336	23.09	MAC	Teratozoospermia	46,XY	N				
102719	27.09	MAC	teratozoospermia	46,XY	N				
102858	27.09	HDE	Infertilidade casal??	46,XY	102860	-	-	-	
103150	29.09	MAC caFertili	infertilidade	46,XY	103152	-	-	-	
103659	01.10	IVI adse	infertilidade	46,XY	103664	-	-	-	
104047	07.10	MAC	Azoospermia	46,XY	104048	não	o	vi	
104055	07.10	MAC	OTA	46,XY	104056	-	-	-	
104150	07.10	MAC	Azoospermia	46,XY	N				
104466	11.10	MAC	oligozoospermia	46,XY	104468	-	-	-	
105250	14.10	MAC	Teratozoospermia	46,XY	N				
108211	02.11	ADSE	Infertilidade	46,XY	108216	-	-	-	
108274	02.11	MAC	OTA	46,XY	108275	-	-	-	
108421	03.11	ARSLVT	Azoospermia	46,XY	108422	-	-	-	
108712	04.11	ARSLVT	Infertilidade casal	46,XY 46,XX	N				
109041	05.11	MAC	OTA	46,XY	N				
110726	16.11	MAC	OTA	46,XY	110727	-	-	-	
111392	22.11	MAC	OTA	46,XY	111394	-	-	-	
112149	25.11	Consultorio . ADMG	infertilidade	46,XY	112150	-	-	-	
112162	25.11	ARSLVT ACESLisboaN	Infert casal	46,XY 46,XX	N				
112476	29.11	MAC	hipogonadismo	46,XY	112478	-	-	-	
112490	29.11	MAC	Azoospermia	46,XY	112491	-	-	-	
112515	29.11	MAC	Azoospermia	46,XY	112516	-	-	-	
112522	29.11	MAC	OTA	46,XY	N				
112815	30.11	MAC	Oligoteratozoospermi	46,XY	N				
113354	06.12	MAC	OTA	46,XY	N				
113644	07.12	MAC	Azoosperm secretória	46,XY	113645	-	-	-	
114184	13.12	MAC	Alteração espermograma	46,XY	N				
114491	16.12	ARSLVT Set CSAmora	OTA	46,XY	114872	-	-	-	
114703	15.12	MAC	Azoospermia	46,XY	N				

114718	15.12	MAC	OTA	46,XY	114719	-	-	-	
114918	16.12	MAC	OTA	46,XY	N				
115399	20.12	MAC	OTA	46,XY	N				
115921	21.12	Consultório?AcesCascais	Infertilidade	46,XY	115922	-	-	-	
115475	20.12	MAC	Infertilidade	46,XY	115476	-	-	-	
116121	22.12	MAC	OTA	46,XY	116122	-	-	-	
116378	23.12	MAC	Azoospermia	46,XY	116380	-	-	-	
116498 X	**27.12**	**MAC**	**OTA**	**47,XYY**	N				
116705	28.12	MAC	Infertilidade	46,XY	N				
116797	28.12	MAC	Infertilidade(OTA)	46,XY	116798	-	-	-	
116964	29.12	MAC	OTA	46,XY	N				
116986	29.12	AvaClinic	Infertilidade	46,XY	116987	-	-	-	
117153	30.12	MAC	OTA	46,XY	117154	-	-	-	
		140???	**11 anomalias** **8 anomalias**	**7.6%** **5.7%**	gr/gr 1	1	3	4	SRY
		Azoospermia Oligozoospermia OTA OT Oligoteratozoo Astenoteratozoospe Teratozoospermia Criptozoospermia Infertilidade Infertilidade casal Não indicado						**1**	
TOTAIS	410		10.2		**16**	**caso**	**com**	**del**	**Y**
					2	**4**	**5**	**12**	**2**
					Mas	**casos**	**com**	**+ de 1**	
Se não entrar	**2015**	**Devido desvirtuar do HGO não indicados**	**Perco 4 transloca e 1 47,XXY**						
383373	12.01	HGO	Não indicado	46,XY,t(8;14)	383374				
386857+	27.01	H Lusíadas	Infertilidade	46,XY,t(1;10)	386858				
399891+	05.03	HDE	Infertilidade	45,rob(13;14)(q10;q10)	399894				
405321+	26.03	ARS Alentejo	Infertilidade	46,XY,t(4;22)(p10;q10)	421323				
406016+	30.03	HDE	Infertilidade	47,XXY	416018				
Totais	**390**	**38-41**			2	4	4	12	2

Casos com cariotipo anormal entre 2010 - Março 2015 (5 anos e 3 meses)

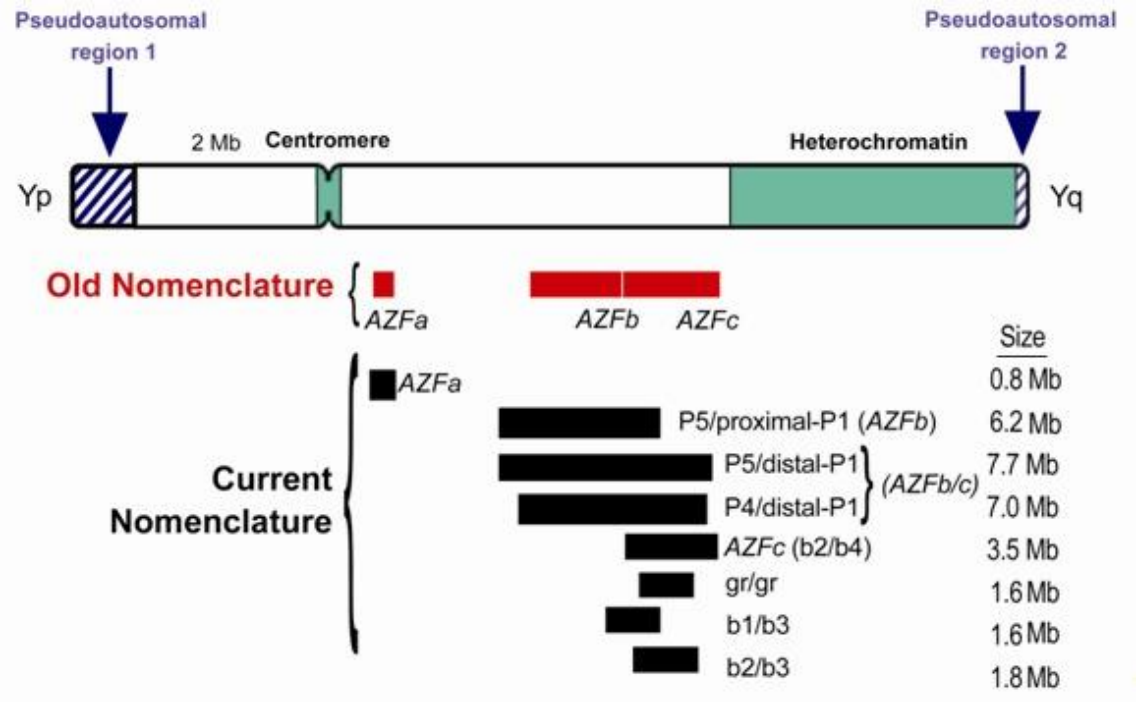
(Resumo)

Klinefelter; translocações; inversões; mosaicismos; marcadores;

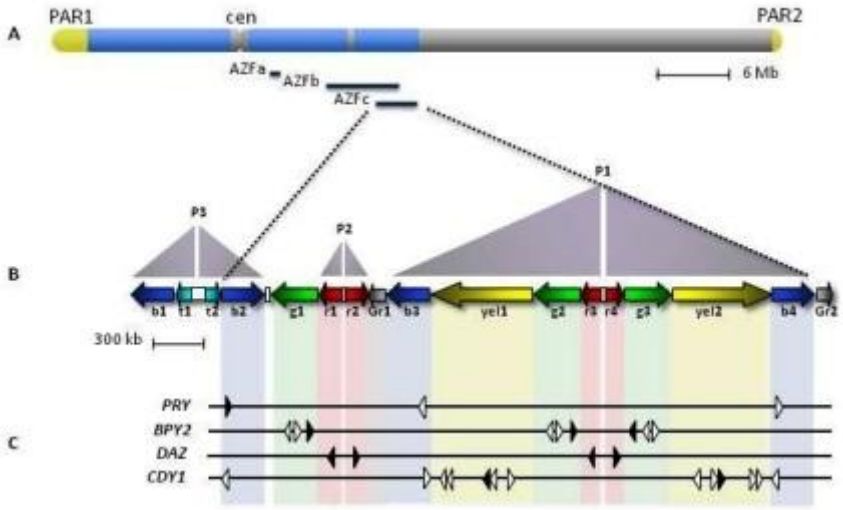
Ano	Requisitante	Indicação	Cariotipo	Estudos Y
		Não indicado	46,XY,t(8;14)	
		Infertilidade	46,XY,t(1;10)	
		Infertilidade	45,rob(13;14)(q10;q10) **SEXO??**	
		Infertilidade	46,XY,t(4;22)(p10;q10)	
		Infertilidade	47,XXY	
		Azoospermia	47,XXY	
		Não indicado	**47,XY,+mar.ISH DER(14/22)(d14z1/d22z1+)**	
		infertilidade	46,XY,t(1;3)(q42.3;q26.2)	

		Não indicado	47,XXY	
		infertilidade	47,XXY	
		azoospermia	**Mos 45,X[34]/ 46,X,idic(Y)(q11.221)[16]**	
		?????????	47,XXY	
		Infertilidade	46,XY,inv(8)???	
		infertilidade	47,XXY	
		Susp S Klinefelter **NÃO ENTRA**	*47,XXY não entra*	
		Não indicado	**47,XXY**	
		Estudo pré-concecional	**46,XY,inv(2)**	
		I	**47,XXY**	
		Não indicado (tb estuY)	47,XXY	
		Infertilidade(OligoAcent	46,XY,t(4;8)(q27; q11.23)	
		Inv(14)(p21.1)(RP11-388M7+)(q21.3)	**(RP11-168D12+), *14q23.3(RP11-430G13x2)***	
		Azoospermia	mos 47,XXY[41]/46,XY[09]	
		Não indicado	47,XXY	
		Não indicado	46,XY,t(7;22)(q11.23;q11.2)	
		OTA oligoastenoteratozoosp	mos 47,XXY[2]/46,XY[48]	
		Azoospermia	46,XY,dup(8)(p23.1)p23.1). ish (wcp8+)	
		Azoospermia	46,X,der(X) ???	
			ishder(X)t(X;Y)(p22.31;p11.2)(SRY+)	
		Não indicado. Desenv sex diminuido NÃO ENTRA	47,XXY	
		Azoospermia	mos 47,XXY[13]/ 46,XX[37]	
		Azoospermia	47,XXY	
		Azoospermia	46,X,+mar,inv(5)	
		(p14.2p15.2)[32]/ 45,X,inv(5)(p14.2p15.2)[18]. Ish i(Y)	(p10)(SRY++,DYZ3+),inv(5)(wcp5+)	**inv + mar + mos**
		Azoospermia	47,XXY	
		Oligozoospermia	**46,XY,rob(13;14)+mar**	**Trans + mar**
		46,XY,rob(13;14)+mar.	**Ish der(16)(wcp16+)**	
		OTA	46,XY,t(4;14;15)	
		Ish der(4)t(4;15)(wcp15+)	,der(14)t(4;14)(wcp4+),der(15)t(14;15)	
		Infer, baixa estatura	**46,X,+mar.ish der(Y)(wcpY+,DYZ3+)**	
		Infertilidade	46,XY,rob(13;14) (q10;q10)	
		SuspeitaKlinefelter **Não entra?**	*Mos 47;XXY[48]/46,XY[2]*	
		Azoospermia por hipogonadismo primário	47,XXY	
		Esposa 2AE Não pôr	46,XY,inv(1)(p13q12) variante	
		Suspeita S Klinefelter **não entra?**	*46,X,der(X)t(X;Y(p22.33;p11.3)*	
		(p22.33;;p11.3). ish der(X)t(X;Y)	*(p22.33;p11.3)(SRY+)*	
		Azoospermia secretória	der(X)t(X;Y(p22.33;p11.3) (wcp	
		(p22.33).ishder(X)t(X;Y)	(p22.33;p11.3)(wcpY+;SRY+)	
		Hipogonadismo primário Susp S. Klinefelter	47,XXY	
		Criptozoospermia	47,XXY	
		Azoospermia. Hipogonadismo primário	47,XXY	
		Azoospermia	47,XXY	
		Azoospermia secretória	47,XXY	
		OTA	**47,XYY**	

		Azoospermia	47,XXY	
		infertilidade	47,XXY	
		Infertilidade; azoospermia	47,XXY	
		Susp S Klinefelter **Não entra?**	*47,XXY*	
		Infertilidade	**47,XYY**	
Totais		**47,XXY** **47,XYY** **trans** **inv** **mar** **mos** **inv mar mos** **der (trans)**	**25** **2** **12 rob:** **5**	



Está no Navarro-Costa??????



Navarro-Costa

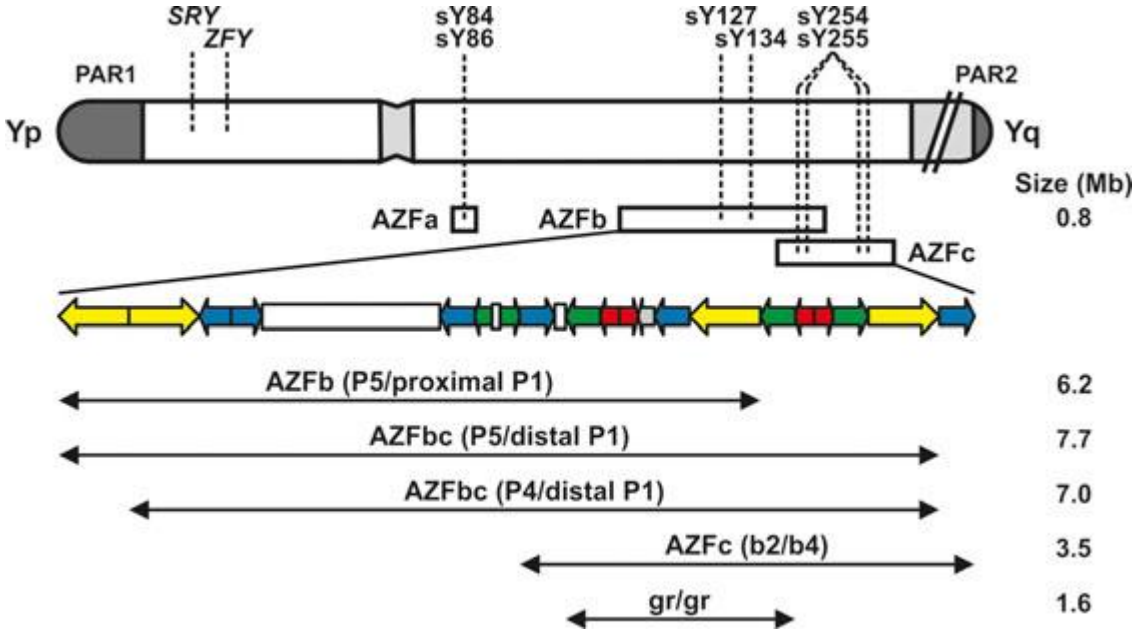


Figure 2 Schematic representation of the Y chromosome and the current microdeletion model (Repping et al., 2002). Repetitive sequences (colour coded palindromes) explain the origin of deletions in the AZFbc region by homologous recombination between identical sequences. The location of the STS primers suggested by the present guidelines is indicated by dashed lines. As four copies of the DAZ gene are normally present on the Y chromosome, the STS primers sY254, sY255 amplify four loci in AZFc. The AZFc (b2/b4) deletion is by far the most frequent type (~80%) of Y-chromosomal microdeletions found

[1]C. Krausz, [2]L. Hoefsloot, [3,4]M. Simoni and [5]F. T€uttelmann "Andrology" Andrology, 2014, 2, 5–19.