**Supplementary Table I. Chromosomal abnormalities in a cohort of 1663 non-iatrogenic azoospermic men**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chromosomal**  **abnormality category** | **Chromosomal abnormality** | **Number of**  **subjects** | **Prevalence** | **Relative proportion of chromosomal abnormalities** | **AZF deletion \*** | **FSH**  **Value** | **Sperm retrieval**  **at surgery** | **Chance of sperm retrieval at surgery** | **Increased**  **risk for**  **miscarriages** | **Increased**  **risk for**  **congenital abnormalities** | **Estimation of**  **risk for**  **congenital abnormalities**  **due to chromosomal abnormality** | **Clinical reference group** |
|  |  | n | n/1663 | n/240 | Relative  proportion | (Mean) value, U/l | Relative  proportion | Relative proportion | Relative proportion | Relative proportion | Absolute risk increase | 0-3 \*\* |
| **Normal**  **karyotype** | **None** | **1423** | **85,6%** | **0%** | **5,4% yes (55/1015)**  **(**39 AZFc deletion)  **94,5% no (960/1015)** | **15,4** | **60% yes (757/1253)**  **40% no (496/1253)** | **100% yes (1423/1423)** | **1423 No (100%)** | **1423 No (100%)** |  | **0** |
| **All chromosomal abnormalities** | **Total** | **240** | **14,4%** | **100%** | **14,7% yes (19/129)**  (2 AZFc deletion)  **85,3% no (110/129)** | **29,9** | **32% yes (64/197)**  **68% no (133/197)** | **93% (224/240)**  **6,7% (16/240)** | **15% (36/240)**  **85% (204/240)** | **12% yes (28/240)**  **88% no (212/240)** |  |  |
| ***Gonosomal,***  ***Klinefelter\*\*\**** | ***Subgroup total*** | ***176*** | ***10,5%*** | ***73,3%*** | ***0% yes (0 /76)*** | ***31,7*** | ***28% yes (42/151)***  ***72% no (109/151)*** | ***100% yes (176/176)*** | ***0% yes (0/176)*** | ***0% yes (0/176)*** |  | ***0*** |
|  | 47,XXY | 167 | 10,0% | 69,6% | 0% Yes (0/72) | 35,3 | 29% yes (41/142)  71% no (101/142) | Yes | No | No |  | 0 |
|  | 47,XXY/46,XY | 9 | 0,5% | 3,8% | 0% Yes (0/4) | 33,0 | 11% yes (1/9)  89% no (8/9) | Yes | No | No |  | 0 |
| ***Gonosomal,***  ***non-Klinefelter*** | ***Subgroup total*** | ***28*** | ***1,7%*** | ***11,7%*** | ***61% yes (17/28)***  *(1 AZF c deletion)*  ***39% no (11/28)*** | ***19,1*** | ***28% yes (4/15)***  ***72% no (11/15)*** | ***43% yes (12/28)***  ***57% no (16/28)*** | ***7,1% yes (2/28)***  ***93% no (26/28)*** | ***7,1% yes (2/28)***  ***93% no (26/28)*** |  |  |
|  | 46,X,der(X)t(X;Y) | 8 | 0,5% | 3,3% | 100% Yes (8/8) | 28,2 | 1 No  7 Unknown | No | No | No |  | 1 |
|  | 46,XX/47,XXY/46,XY | 1 | 0,06% | 0,4% | No | 34 | No | Yes | No | No |  | 0 |
|  | 47,XYY/46,XY | 1 | 0,06% | 0,4% | No | 5,2 | No | Yes | No | No |  | 0 |
|  | 47,XYY/46,XY | 1 | 0,06% | 0,4% | No | 8,6 | Yes | Yes | No | No |  | 0 |
|  | 45,X/46,X,der(Y) | 1 | 0,06% | 0,4% | Yes | 24,3 | Unknown | No | No | No |  | 1 |
|  | 45,X/46,X,der(Y) | 1 | 0,06% | 0,4% | No | 13,2 | Unknown | Yes | No | No |  | 0 |
|  | 45,X/46,X,r(Y) | 1 | 0,06% | 0,4% | No | 7,0 | Unknown | Yes | Yes | Yes | Early loss r(Y): Turner syndrome | 2,3 |
|  | 45,X/46,X,r(Y) | 1 | 0,06% | 0,4% | No | 4,9 | Yes | Yes | Yes | Yes | Early loss r(Y): Turner syndrome | 2,3 |
|  | 45,X/46,X.idic(Yq12) | 1 | 0,06% | 0,4% | No | 22 | Yes | Yes | No | No |  | 0 |
|  | 45,X/46,XY | 1 | 0,06% | 0,4% | No | 14,2 | Unknown | Yes | No | No |  | 0 |
|  | 45,X/46,XY | 1 | 0,06% | 0,4% | No | 16 | No | Yes | No | No |  | 0 |
|  | 45,X/46,XY | 1 | 0,06% | 0,4% | No | 6 | Yes | Yes | No | No |  | 0 |
|  | 46,X,del(Y)(q11) | 1 | 0,06% | 0,4% | Yes | 28,0 | Unknown | No | No | No |  | 1 |
|  | 46,X,del(Y)(q11.2) | 1 | 0,06% | 0,4% | Yes (AZFc deletion) | 15,3 | No | Yes | No | No |  | 0 |
|  | 46,X,del(Y)(q11.2) | 1 | 0,06% | 0,4% | Yes | 27,4 | No | No | No | No |  | 1 |
|  | 46,X,del(Y)(q11.2) | 1 | 0,06% | 0,4% | Yes | 18 | No | No | No | No |  | 1 |
|  | 46,X,der(Y) | 1 | 0,06% | 0,4% | Yes | 7 | No | No | No | No |  | 1 |
|  | 46,X,idic(Y)(q11) | 1 | 0,06% | 0,4% | Yes | 9 | No | No | No | No |  | 1 |
|  | 46,X,idic(Y)(q11.21) | 1 | 0,06% | 0,4% | Yes | 16,3 | No | No | No | No |  | 1 |
|  | 46,X,r(Y)(p11.32q12) | 1 | 0,06% | 0,4% | Yes | 8 | Unknown | No | No | No |  | 1 |
|  | 46,XY/46,X,del(Y)(q?) | 1 | 0,06% | 0,4% | No | 24,3 | No | Yes | No | No |  | 0 |
| ***Translocation, autosomal*** | ***Subgroup total*** | ***12*** | ***0,7%*** | ***5,0%*** | ***0% Yes (0/10)*** | ***10,7*** | ***45% yes (5/11)***  ***55% no (6/11)*** | ***100% yes (12/12)*** | ***100% yes (12/12)*** | ***67% yes (8/12)***  ***33% no (4/12)*** |  |  |
| ***reciprocal*** | 46,XY,t(1;6)(pter;q24.2) | 1 | 0,06% | 0,4% | Unknown | 4 | No | Yes | Yes | Yes | <<1% | 2,3 |
|  | 46XY,t(1,10)(p36;q22) | 1 | 0,06% | 0,4% | No | 11,2 | No | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,XY,t(1;13)(p10;q10) | 1 | 0,06% | 0,4% | No | 11,5 | No | Yes | Yes | No |  | 2 |
|  | 46,XY,t(1;14)(p10;q10) | 1 | 0,06% | 0,4% | No | 14 | No | Yes | Yes | Yes | UPD14: <1% | 2,3 |
|  | 46,XY,t(1;17)(p36.3;q11.2) | 1 | 0,06% | 0,4% | No | 11,9 | Yes | Yes | Yes | No |  | 2 |
|  | 46,XY,t(3;7)(q25.3?;p14.2?) | 1 | 0,06% | 0,4% | No | 2 | Yes | Yes | Yes | No |  | 2 |
|  | 46,XY,t(4;13)(q35;q12.3) | 1 | 0,06% | 0,4% | No | 12 | Yes | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,XY,t(5;14)(q35.3?;q24;1?) | 1 | 0,06% | 0,4% | Unknown | 4,4 | No | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,XY,t(6;15)(q13?;q22?) | 1 | 0,06% | 0,4% | No | 10,7 | Unknown | Yes | Yes | No |  | 2 |
|  | 46,XY,t(11;14)(p10;q11.2) | 1 | 0,06% | 0,4% | No | 5,9 | Yes | Yes | Yes | Yes | UPD14: <1% | 2,3 |
|  | 46,XY,t(16;21)(q11.2;p11.2) | 1 | 0,06% | 0,4% | No | 4,7 | No | Yes | Yes | Yes | 1% | 2,3 |
|  | 46,XY,t(18;19 (p11.2;p11) | 1 | 0,06% | 0,4% | No | 3,3 | Yes | Yes | Yes | Yes | <1% | 2,3 |
| ***Translocation,***  ***autosomal Robersonian*** | ***Subgroup total*** | ***11*** | ***0,7%*** | ***4,6%*** | ***33% yes (2/6)***  *(1 AZFc deletion)*  ***67% no (4/6)*** | ***45,7*** | ***70% yes (7/10)***  ***30% no (3/10)*** | ***100% yes (11/11)*** | ***100% yes (11/11)*** | ***100% yes (11/11)*** |  | ***2,3*** |
|  | 45,XY,rob(13;14)(q10;q10) | 6 | 0,4% | 2,5% | 50% yes (2/4)  (1 AZFc deletion)  50% no (2/4) | 20,2 | 4 Yes  2 No | Yes | Yes | Yes | Trisomie 13: 1%, UPD14: 0.5% | 2,3 |
|  | 45,XY,rob(13;15)(q10;q10) | 1 | 0,06% | 0,4% | No | 5 | Yes | Yes | Yes | Yes | Trisomie 13: 1%, UPD15: 0.5% | 2,3 |
|  | 45,XY,rob(13;15)(q10;q10) | 1 | 0,06% | 0,4% | Unknown | 45,7 | Unknown | Yes | Yes | Yes | Trisomie 13: 1%, UPD15: 0.5% | 2,3 |
|  | 45,XY,rob(14;15)(q10;q10) | 1 | 0,06% | 0,4% | Unknown | 16,8 | Yes | Yes | Yes | Yes | UPD14: 0.5%, UPD15: 0.5% | 2,3 |
|  | 45,XY,rob(14;15)(q10;q10) | 1 | 0,06% | 0,4% | No | 18 | Yes | Yes | Yes | Yes | UPD14: 0.5%, UPD15: 0.5% | 2,3 |
|  | 45,XY,rob(14;21)(q10;q10) | 1 | 0,06% | 0,4% | Unknown | 16 | No | Yes | Yes | Yes | Trisomie 21: 1-5%, UPD14: 0.5% | 2,3 |
| ***Translocation,***  ***involving*** | ***Subgroup total*** | ***9*** | ***0,5%*** | ***3,8%*** | ***0% yes (0/6)*** | ***9,0*** | ***43% yes (3/7)***  ***57% no (4/7)*** | ***100% yes (9/9)*** | ***89% yes (8/9)***  ***11% no (1/9)*** | ***67% yes (6/9)***  ***33% no (3/9)*** |  |  |
| ***gonosomes*** | 46,Y,t(X;1)(p11.4;q42.1) | 1 | 0,06% | 0,4% | No | 3,7 | No | Yes | Yes | Yes | 5-10% | 2,3 |
|  | 46,Y,t(X;7)(q26.1;q21.3) | 1 | 0,06% | 0,4% | No | 8,3 | Unknown | Yes | No | Yes | 1% | 3 |
|  | 46,Y,t(X;15)(q26.1;q24.1) | 1 | 0,06% | 0,4% | Unknown | 4,9 | Yes | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,Y,t(X;15)(q26.1;q24.1) | 1 | 0,06% | 0,4% | Unknown | 5,6 | Yes | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,X,t(Y;1)(q11.2;q12) | 1 | 0,06% | 0,4% | No | 20 | No | Yes | Yes | No |  | 2 |
|  | 46,X,t(Y;2)(q10?;q10?) | 1 | 0,06% | 0,4% | Unknown | 5,1 | Yes | Yes | Yes | No |  | 2 |
|  | 46X,t(Y;7)(q11;p22) | 1 | 0,06% | 0,4% | No | 4 | No | Yes | Yes | No |  | 2 |
|  | 45,X,der(Y)t(Y;13)(q10;q10) | 1 | 0,06% | 0,4% | No | 20 | No | Yes | Yes | Yes | 1-5% | 2,3 |
|  | 46,X,t(Y;16)(q11.22;p13.2) | 1 | 0,06% | 0,4% | No | 9,4 | Unknown | Yes | Yes | Yes | 1-5% | 2,3 |
| ***Remaining***  ***category*** | ***Subgroup total*** | ***4*** | ***0,2%*** | ***1,7%*** | ***0% yes (0/3)*** | ***8,0*** | ***100% yes (3/3)*** | ***100% yes (4/4)*** | ***75% yes (3/4)***  ***25% no (1/4)*** | ***25% yes (1/4)***  ***75% no (3/4)*** |  |  |
| Inversion, autosomal | 46,XY,inv(10)(p13q23.1) | 1 | 0,06% | 0,4% | Unknown | 5 | Yes | Yes | Yes | Yes | <1% | 2,3 |
| Inversion, autosomal | 46,XY,inv(7)(p13?q22?) | 1 | 0,06% | 0,4% | No | 13 | Yes | Yes | Yes | No |  | 2 |
| Ring chromosome, autosomal | 46,XY,r(22)(p?;q?) | 1 | 0,06% | 0,4% | No | 9,7 | Unknown | Yes | Yes | No |  | 2 |
| Marker chromosome, autosomal | 47,XY,+idic(15)(q?) | 1 | 0,06% | 0,4% | No | 4,3 | Yes | Yes | No | No |  | 0 |

>5 identical chromosomal abnormalities are combined

\*AZF deletions comprise all (combinations of) AZF deletions except isolated AZFc deletions (described separately)

\*\*Clinical reference group: 0=no clinical relevance; 1=absence of spermatogenesis; 2=increased risk for miscarriage; 3=increased risk for congenital abnormality

\*\*\*Mosaic and non-mosaic Klinefelter syndrome