

**Repping et al., Polymorphism for a 1.6-Mb deletion of the human Y chromosome persists through balance between recurrent mutation and haploid selection**

**Supplementary Table 1.** Men with STS results indicating likely gr/gr deletion: they lacked sY1291 but possessed sY1161, sY1191, sY1206, and sY1201 (**Fig. 1a**, main text).

Sample ID	Branch of Y genealogy <sup>a</sup>	# red (cosmid 18E8) signals per Y chromosome by FISH	Sequence of green (G) and red (R) signals ( <b>Figs. 1 and 3</b> , main text)
<b>Initial screen, spermatogenic failure</b>			
WHT2709	F*(xH,I,J,K)	1	G-R-G
WHT3107	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
WHT3173	R1*(xR1a,R1/-USP9Y+3636)	2	G-R-G-G-R-G
WHT3315	J*(xJ2)	1	G-R-G
WHT3390	J*(xJ2)	1	G-R-G
WHT3393	J*(xJ2)	cells unavailable	
WHT3464	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
WHT3574	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
WHT3662	I	1	G-R-G
WHT3890	J2e	1	G-R-G
WHT3974	E3b2	1	G-R-G
WHT3250	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
WHT3338	K/-USP9Y+3178	1	G-R-G
WHT3674	E3a	1	G-R-G
WHT3795	F*(xH,I,J,K)	1	G-R-G
WHT3829	F*(xH,I,J,K)	1	G-R-G
WHT3975	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
<b>Initial screen, unknown spermatogenic phenotype</b>			
GM04535	D2b	1	G-R-G
PD0045	O3*(xO3e)	1	G-R-G
PD0178	D2b	1	G-R-G
PD0296	Q3	2	G-R-G-G-R-G
PD0373	J2*(xJ2e,J2f)	cells unavailable	
<b>Association study, spermatogenic failure</b>			
AMC0006	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
AMC0073	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
AMC0101	E3b3	1	G-R-G
AMC0102	E3b2	1	G-R-G
AMC0103	R1*(xR1a,R1/-USP9Y+3636)	cells unavailable	
AMC0104	R1*(xR1a,R1/-USP9Y+3636)	1	insufficient cells for additional FISH analysis
AMC0105 <sup>b</sup>	R1*(xR1a,R1/-USP9Y+3636)	2 <sup>b</sup>	insufficient cells for additional FISH analysis
AMC0106	R1*(xR1a,R1/-USP9Y+3636)	1	G-R-G
AMC0107	E3a	1	G-R-G
AMC0108	R1a	cells unavailable	
<b>Additional branch D2b samples</b>			
AG10646	D2b	cells unavailable	
GM00743	D2b	1	G-R-G
GM10810	D2b	1	G-R-G
PD0436	D2b	1	G-R-G
TDC0001	D2b	cells unavailable	
TDC0002	D2b	cells unavailable	
TDC0003	D2b	cells unavailable	
TDC0004	D2b	cells unavailable	
TDC0005	D2b	cells unavailable	
WHT3341	D2b	1	G-R-G

<sup>a</sup> See **Supplementary Figure 1**; nomenclature from the Y Chromosome Consortium (*Genome Res.* **12**, 339-348, 2002).

<sup>b</sup> Excluded from association study because of 47,XYY karyotype and duplication within *AZFc* (**Fig. 3**, main text). See **Supplementary Note 5** for alternative analysis.