

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 6. References for polymorphisms defining the Y haplotypes and genealogy in **Figure 4** (main text), **Supplementary Tables 1–4**, **Supplementary Table 7** and **Supplementary Figure 1**.

Polymorphism	Reference	Polymorphism	Reference
DYS257	1	M50	5
DYS271 (M2)	2	M51	3
M109	3	M58	3
M112	3	M60	5
M116	3	M64	5
M117	3	M67	5
M118	3	M69	5
M119	3	M75	5
M12	4	M76	3
M122	3	M78	3
M123	3	M81	5
M124	3	M82	5
M13	4	M89	5
M134	3	M9	4
M14	4	M91	5
M144	5	M92	5
M168	5	M95	5
M170	5	M96	3
M172	5	p12f	7, assayed as in ref. 8
M173	5	RPS4Y711	9
M175	5	Tat	10
M20	4	SRY10831	11
M3	6	USP9Y+3178	12
M32	3	USP9Y+3636	12
M35	3	YAP	13
M4	4		

Supplementary Table 6 References

1. Hammer, M.F. *et al.* Out of Africa and back again: nested cladistic analysis of human Y chromosome variation. *Mol. Biol. Evol.* 15, 427-41 (1998).
2. Seielstad, M.T. *et al.* Construction of human Y-chromosomal haplotypes using a new polymorphic A to G transition. *Hum. Mol. Genet.* 3, 2159–2161 (1994).
3. Underhill, P.A. *et al.* Y chromosome sequence variation and the history of human populations. *Nature Genet.* 26, 358-361 (2000).
4. Underhill, P.A. *et al.* Detection of numerous Y chromosome biallelic polymorphisms by denaturing high-performance liquid chromatography. *Genome Res.* 7, 996-1005 (1997).

5. Shen, P. *et al.* Population genetic implications from sequence variation in four Y chromosome genes. *Proc. Natl. Acad. Sci. U. S. A.* 97, 7354-7359 (2000).
6. Underhill, P.A., Jin, L., Zemans, R., Oefner, P.J. & Cavalli-Sforza, L.L. A pre-Columbian Y chromosome-specific transition and its implications for human evolutionary history. *Proc. Natl. Acad. Sci. U. S. A.* 93, 196-200 (1996).
7. Casanova, M. *et al.* A human Y-linked DNA polymorphism and its potential for estimating genetic and evolutionary distance. *Science* 230, 1403-1406 (1985).
8. Sun, C. *et al.* Deletion of azoospermia factor a (AZFa) region of human Y chromosome caused by recombination between HERV15 proviruses. *Hum. Mol. Genet.* 9, 2291-2296 (2000).
9. Bergen, A.W. *et al.* An Asian-Native American paternal lineage identified by *RPS4Y* resequencing and by microsatellite typing. *Ann. Hum. Genet.* 63, 63-80 (1999).
10. Zerjal, T. *et al.* Genetic relationships of Asians and Northern Europeans, revealed by Y-chromosomal DNA analysis. *Am. J. Hum. Genet.* 60, 1174-1183 (1997).
11. Whitfield, L.S., Lovell-Badge, R. & Goodfellow, P.N. Rapid sequence evolution of the mammalian sex-determining gene *SRY*. *Nature* 364, 713-715 (1993).
12. Sun, C. *et al.* An azoospermic man with a *de novo* point mutation in the Y-chromosomal gene *USP9Y*. *Nature Genet.* 23, 429-432 (1999).
13. Hammer, M.F. A recent insertion of an Alu element on the Y chromosome is a useful marker for human population studies. *Mol. Biol. Evol.* 11, 749-761 (1994).