

Supplemental Data

An African American Paternal Lineage

Adds an Extremely Ancient Root to the

Human Y Chromosome Phylogenetic Tree

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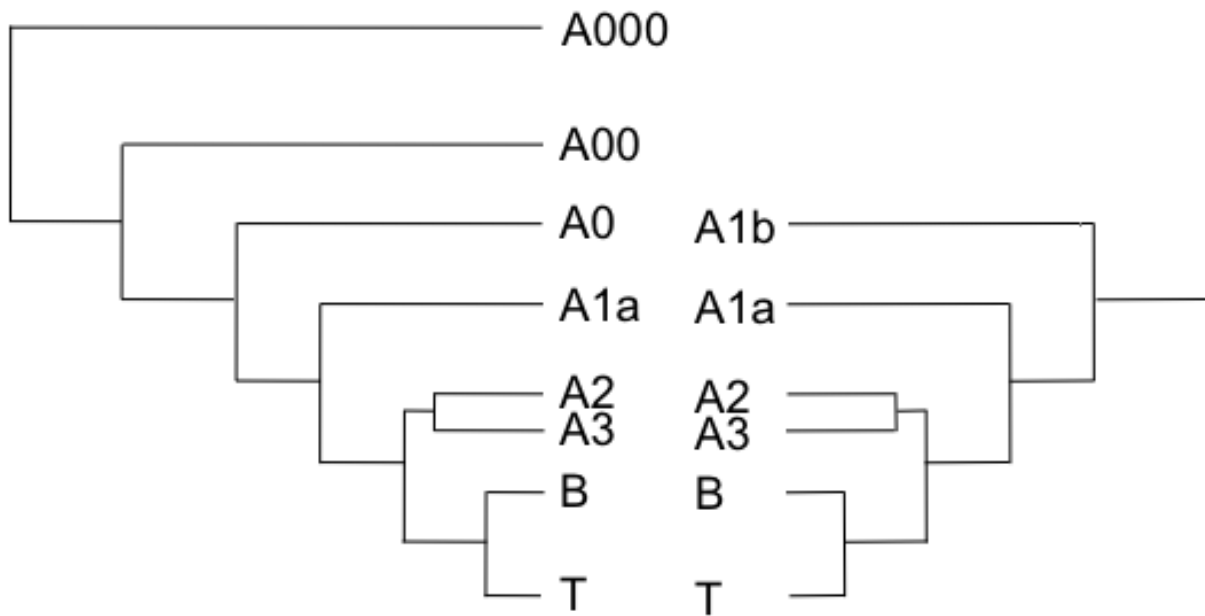


Figure S1. Proposed Nomenclature System for Newly Discovered Deep Branches (Those Branches that Carry the Ancestral State at All Y-SNPs that Were Previously Known)

On the left is shown the proposed nomenclature (see text), and on the right is shown the nomenclature of Cruciani et al.¹. We have renamed the basal branch in Cruciani et al.¹ as A0 (previously A1b) and refer to the presently reported lineage as A00. For deep branches discovered in the future, we suggest continuing the nomenclature A000, and so on. If multiple deep branches are found, then a possible short hand version could be used, such as A(0)n.

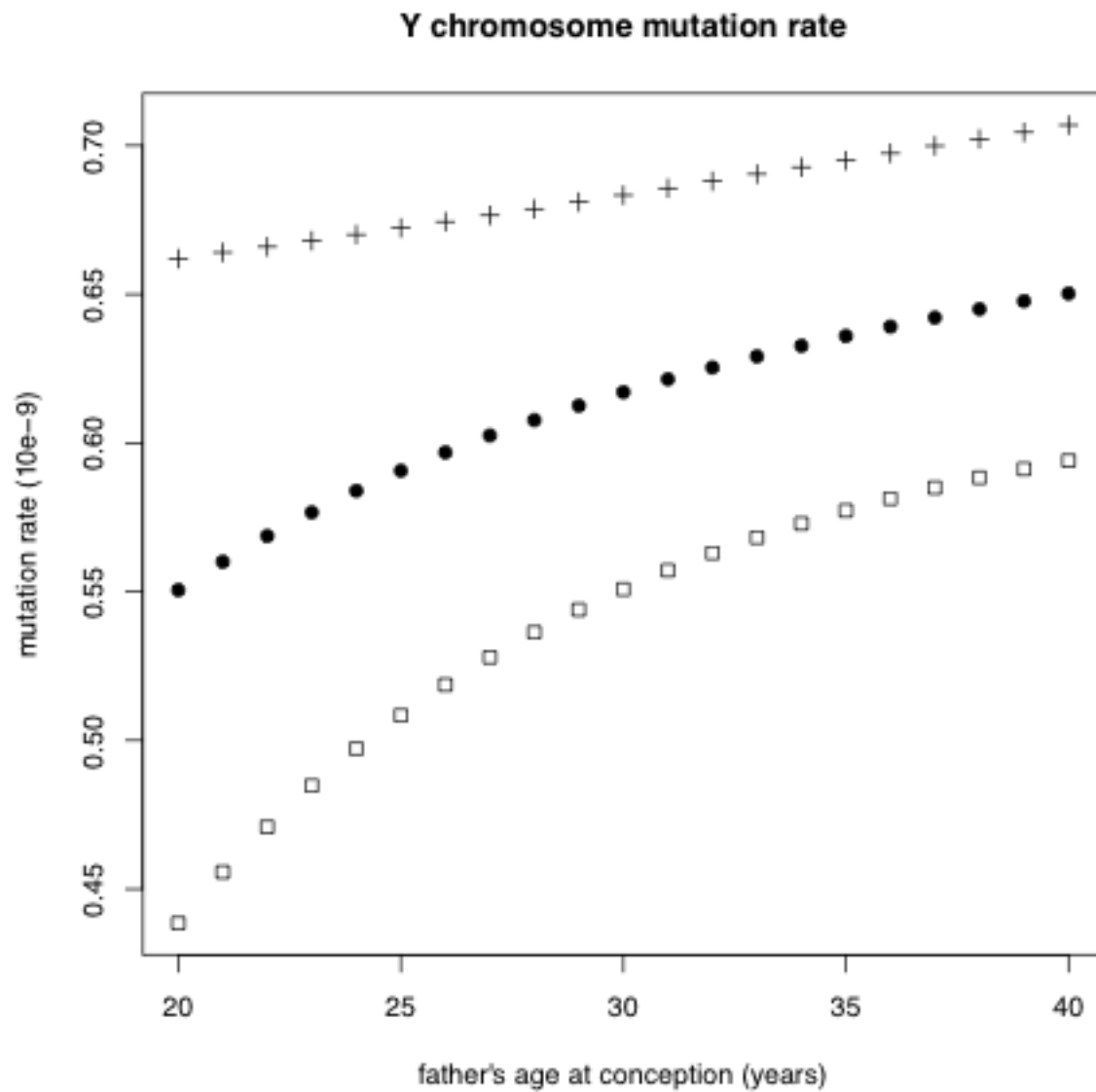


Figure S2. Estimates for the X-Degenerate Y Chromosome Mutation Rate per Year per Base, as Inferred from Mutation Rates Estimated in Pedigrees

For each age of reproduction, the lower bound (open squares '□'), median (solid circles '●') and upper bound (plus signs '+') are indicated.

Table S1. Mutations in A00 and A0 Chromosomes

Position Start (hg18)	Position End (hg18)	Branch	Name	Mutation	Notes
2716961	2716961	A00	L1234	G->A	+ in Mbo
2838396	2838396	A00	L1122	C->T	+ in Mbo
2886312	2886312	A00	L1086	A->T	+ in Mbo
2935499	2935499	A00	L1087	G->T	+ in Mbo
2947279	2947279	A00	L1088	C->G	+ in Mbo
2947280	2947280	A0-T	L1089	G->C	
6895384	6895384	A00	L1096	G->A	+ in Mbo
6903217	6903217	A00	L1097	C->T	+ in Mbo
6911661	6911661	A00	L1100	C->T	- in Mbo
6936455	6936455	A00	L1102	A->G	+ in Mbo
6937771	6937771	A00	AF6	A->G	+ in Mbo
6939913	6939913	A00	L1103	C->T	+ in Mbo
6955758	6955758	A00	L1104	C->T	+ in Mbo
6991670	6991670	A0	L1073	T->C	
6991719	6991719	A00	AF7	A->G	+ in Mbo
7121505	7121505	A0	L984	G->T	
12511289	12511289	A00	L990	A->G	
12635645	12635645	A0	L1075	C->T	
12636859	12636859	A00	AF8	A->G	+ in Mbo
12741291	12741291	A0-T	L1118	T->C	
12796426	12796426	A00	L1117	A->T	+ in Mbo
12917922	12917922	A00	L1119	G->A	+ in Mbo
12979380	12979380	A0	L993	A->C	
13006456	13006456	A0-T	L1121	G->A	
13007067	13007067	A0	L991	C->A	
13414541	13414541	A0	L995	T->C	
13533434	13533434	A0	L996	G->A	
13935738	13935738	A0-T	L1124	T->C	
13946200	13946200	A0	L998	T->G	
13947281	13947281	A0	L999	C->T	
13977162	13977162	A0-T	L1125	A->G	
14084737	14084737	A0	L1000	C->A	
14114397	14114397	A00	L1126	A->G	+ in Mbo
14431477	14431477	A0	L1076	G->A	
14702402	14702402	A00	AF9	C->T	+ in Mbo
14791204	14791204	A0-T	L1127	A->G	
14838487	14838487	A00		G->A	
14860990	14860990	A00	AF10	G->A	+ in Mbo
14901994	14901994	A0	L1001	G->T	
14902035	14902035	A0-T	L1273	T->C	
15106240	15106240	A0-T	L1129	T->C	used gorilla to infer ancestral state (6 bp deletion in chimp)
15170404	15170404	A0-T	L1130	T->G	
15208490	15208490	A00	L1131	G->A	+ in Mbo
15228205	15228205	A0-T	L1132	A->G	
15695829	15695829	A00	L1133	C->T	+ in Mbo
15976206	15976206	A0	L1077	G->T	

Table S1. Continued

Position Start (hg18)	Position End (hg18)	Branch	Name	Mutation	Notes
16019965	16019965	A00	L1134	G->T	+ in Mbo
16283616	16283616	A0	L1006	A->T	
16627408	16627408	A0	L1008	C->A	
16656697	16656697	A0-T	L1135	C->A	
17374735	17374736	A0	L1078	CC->AT	
17423680	17423680	A0-T	L1136	A->G	
17556485	17556485	A0-T	L1137	C->T	
17558114	17558114	A00	L1138	G->T	+ in Mbo
17587144	17587144	A00	L1139	A->C	+ in Mbo
17885711	17885711	A00	L1140	G->C	+ in Mbo
19502698	19502698	A00	L1141	C->T	+ in Mbo
19502834	19502834	A0	L1010	T->C	
19747950	19747950	A0-T	L1142	C->T	
19748116	19748116	A0	L1011	C->A	
19940817	19940817	A0	L1012	C->T	
19950468	19950468	A00	AF13	G->A	+ in Mbo
19966682	19966682	A00		G->T	
20052733	20052733	A0-T	L1143	A->G	
20081484	20081484	A0	L1055	G->A	
20166014	20166014	A0	L896	A->T	
20176694	20176694	A00	L1144	C->T	+ in Mbo
20199048	20199048	A00	L1146	G->A	+ in Mbo
20199178	20199178	A0-T	L1145	C->T	
20211928	20211928	A00	L1147	A->T	+ in Mbo
20328703	20328703	A00	L1148	C->T	+ in Mbo
20329216	20329216	A00	L1149	C->T	- in Mbo
20349061	20349061	A0	L1015	G->A	
20352688	20352688	A0	L1080	T->C	
20355437	20355437	A0	L1016	T->C	
20363359	20363359	A0-T	L1150	A->G	
20376381	20376381	A00	L1151	G->T	+ in Mbo
20383724	20383724	A0	L1017	C->G	
20385338	20385338	A00	L1152	C->T	+ in Mbo
20638051	20638051	A0	L1070	G->C	
20650519	20650519	A00	L1154	A->G	+ in Mbo
20650654	20650654	A0-T	L1155	G->C	
21132180	21132180	A0	L1083	G->T	
21378369	21378369	A00	L1156	C->G	+ in Mbo
21386451	21386451	A00	L1157	G->A	+ in Mbo
21419449	21419449	A00	L1158	G->T	
21431054	21431054	A00	L1159	A->T	+ in Mbo
21527318	21527318	A00	L1160	A->G	+ in Mbo

Table S2. Continued

Y-STR	Sample											
	Mbo052	Mbo159	Mbo160	Mbo161	Mbo170	Mbo173	Mbo174	Mbo179	Mbo183	Mbo186	Mbo199	African American
DYS715	26	26	26	26	26	26	26	26	26	26	26	26
DYS716	28	28	28	-1	28	28	28	-1	28	28	28	28
DYS717	19	19	19	19	19	19	19	-1	19	19	19	19
DYS726	15	15	15	15	15	15	15	-1	15	15	15	15
Y-GATA-A10	12	12	12	11	12	12	12	12	12	12	12	12
Y-GATA-H4	10	10	10	10	10	10	10	10	10	10	10	10
Y-GGAAT-1B07	10	10	10	10	10	10	10	10	10	10	10	10

^a: From a pool of 102 STRs, multiple copy STRs with undistinguishable copies (CDY, DYF395S1, DYS385, DYS413, DYS459, DYS464, and YCAII) were removed.

Data from three individuals lacking information for 8 or more STRs were removed from further analyses.

^b: No data is indicated with -1

^c: all listed samples have a .2 micro allele

^d: all listed samples have a .3 micro allele

^e: Mbo have a .2 micro allele and the African American individual has a .1 micro allele

^f: all listed samples have a .3 micro allele

Reference

1. Cruciani, F., Trombetta, B., Massaia, A., Destro-Bisol, G., Sellitto, D., and Scozzari, R. (2011). A revised root for the human Y chromosomal phylogenetic tree: the origin of patrilineal diversity in Africa. *Am J Hum Genet* 88, 814-818.