

Fig. 4a Y-DNA111 Mutation Frequencies

Introduction

When constructing our set of McCarthy Surname Study phylogenetic trees in the absence of SNP information such as is provided with Big Y testing, identification of groups of shared Y-STR mutations is usually more important than Genetic Difference. If STR loci are known to mutate very rarely, these on their own or in conjunction with just a small number of other STR mutations may point to a shared ancestry. Even when SNP data is available, study of STR mutations can often point to Big Y results sets to be searched more thoroughly e.g. with the use of BAM files for potential confirmatory SNPs which FTDNA's own analysis has rejected for not meeting its acceptance criteria. So it is useful to know the frequency with which STR markers have been found to mutate, particularly for the slower mutating loci.

Various analyses and papers have been presented over the years with estimates of mutation rates for the 111 loci of FTDNA's Y-DNA111 test. Many break the numbers down into groups, e.g. x, slow, medium, fast, x.fast. The McCarthy Study has hitherto used an average of these, assigning a mutation rate category, VS, S, M, F, VF respectively in its table of McCarthy Ancestral Haplotypes (Fig. 4, on this Wordpress site). On the McCarthy Study trees the slow ('S') mutating STR loci are colour highlighted yellow, while the very slowest ('VS') are highlighted lime green.

However, with ten years' experience of placement of STR mutations on McCarthy and other phylogenetic trees mostly serving the haplogroup of SNP R-L21, it has become evident that some revisions are warranted to the rates used as a basis for these categorisations. Counts of over 5,000 mutation occurrences made at the end of January 2020 on the Irish Type II tree (which also serves McCarthy R-L21 Group A) and the R-Z16526 tree (which also serves McCarthy R-L21 Group B) have been weighted in consideration of the varying number of markers tested by different participants (normally 37, 67 or 111, occasionally 25, 43). The combined count has then been similarly weighted in favour of the Irish Type II tree (935 tests vs 300 for the Z16526 tree). The revised mutation frequency categorisations for each of the 111 markers of FTDNA's Y-DNA111 test are indicated in the table below, alongside those used hitherto. The cut-off points between the categories are arbitrary and should not be compared with those made by other analysts. Revisions to colour highlighting for Very Slow and Slow mutating markers are now shown on all the Study's trees. It is important to appreciate the following:

- The table does NOT offer actual mutation frequencies. The 'weighted frequencies' indicate the relative propensity of the various STR loci to mutate based on the counts described above.

- Susceptibility to mutation and direction of that mutation at any STR site may be dependent on the allele prevailing. For instance it is very clear that DYS442 is far more likely to mutate when its allele is 13 than when 12 (its status at the time of SNP R-L21), and then a mutation back to 12 is more likely than one forward to 14 (See also Note 2 below the table).

- Where the trees show multi-step mutations, these have been counted as single events. This appears to be valid for the mutations from 22 to 18 at DYS481 in the haplogroup of SNP A212, and of 23 to 21 or 19 at DYS413a but may not be so where, for example, a 38 to 36 mutation is indicated at CDYb. Any errors arising therefrom will not affect the categorisations.

STR Name	FTDNA listing order of appearance	Order of Mutation frequency (slowest to fastest)	Weighted Frequency (see Introduction)	Mutation Rate Category		Notes
				Old	New (from Feb 2020)	
DYS590	42	1	0.0	VS	VS	
DYS472	45	1	0.0	VS	VS	
DYS490	54	3	0.8	VS	VS	
DYS450	56	4	1.4	VS	VS	
DYS426	7	5	1.5	VS	VS	
DYS436	53	5	1.5	VS	VS	
DYS435	111	5	1.5	VS	VS	
DYS575	84	8	2.0	VS	VS	
DYS726	103	8	2.0	VS	VS	
DYS459a	14	10	2.2	S	VS	
DYS494	81	11	2.5	VS	VS	
DYS632	70	12	2.9	VS	VS	
DYS640	65	13	3.0	VS	VS	
DYS454	17	14	3.2	VS	VS	
DYS568	62	15	3.4	VS	VS	
DYS593	95	16	3.5	VS	VS	
DYS617	61	17	3.6	VS	VS	
DYS425	48	18	4.1	VS	VS	
DYS388	8	19	4.6	VS	VS	
DYS455	16	20	5.1	VS	VS	
DYS492	66	21	5.2	VS	VS	
DYS578	39	22	5.3	VS	VS	
DYF395S1b	41	23	5.4	VS	VS	
DYS531	38	24	5.6	VS	VS	
DYS636	83	25	6.0	S	VS	
DYS641	44	26	6.4	VS	VS	
DYS434	109	27	6.5	VS	VS	
YCAIIa	28	28	6.8	S	VS	
DYS594	52	29	7.2	VS	VS	
DYS438	37	30	7.8	S	S	
DYS556	77	30	7.8	S	S	
DYS717	75	32	8.4	M	S	
DYS462	86	33	8.8	S	S	
DYS638	85	34	8.9	S	S	
DYF395S1a	40	35	9.2	VS	S	
Y-GGAA1-TB07	92	36	9.3	S	S	
DYS537	43	37	9.4	S	S	
DYS565	67	37	9.4	S	S	
DYS393	1	39	10.9	S	S	
DYS392	11	39	10.9	S	S	
DYS589	79	41	12.4	S	S	
DYS463	90	42	12.9	S	S	
DYS385a	5	42	12.9	M	S	
DYS445	88	44	13.8	S	S	
DYS540	72	44	13.8	S	S	
DYS448	20	44	13.8	S	S	
DYS487	63	47	14.5	S	S	
DYS716	74	48	15.3	VS	S	
DYS441	91	49	15.7	M	S	
DYS643	106	50	15.8	S	S	
DYS437	19	50	15.8	S	S	
DYS495	71	52	16.3	S	S	
DYF406S1	46	53	17.6	S	S	
DYS572	64	53	17.6	M	S	
DYS561	101	55	18.0	S	S	
DYS497	107	56	19.1	S	S	
YCAIIb	29	57	19.9	S	S	
DYS587	105	58	20.3	S	S	
DYS19/394	3	59	20.4	S	S	
DYS391	4	60	20.6	M	S	
DYS522	80	61	20.7	S	S	
DYS520	59	62	20.9	M	S	
DYS459b	15	63	21.1	S	S	
DYS461	110	64	21.2	M	S	
DYS389-2 - DYS389-1	12	65	22.1	S	S	1
DYS525	93	66	22.2	S	S	
DYS511	47	67	23.0	S	S	
DYS485	69	68	23.1	S	S	
Y-GATA-H4	27	69	26.0	M	M	
DYS452	87	70	26.2	M	M	
DYS464a	22	71	26.5	M	M	
DYS464b	23	71	26.5	M	M	
DYS413a	49	73	27.6	S	M	
DYS413b	50	73	27.6	S	M	
DYS505	76	75	28.1	M	M	
DYS607	31	76	29.4	M	M	
DYS389-1	10	77	34.0	S	M	
DYS456	30	78	36.5	F	M	
DYS552	102	79	41.1	M	M	
DYS439	9	80	42.0	M	M	
DYS557	51	81	42.4	M	M	
DYS513	100	82	42.5	M	M	
DYS481	58	83	42.9	M	M	
DYS390	2	84	44.0	M	M	
DYS447	18	85	45.2	M	M	
DYS444	57	86	45.3	M	M	
DYS635	104	87	46.2	M	M	
DYS464c	24	88	46.5	M	M	
DYS464d	25	88	46.5	M	M	
DYS385b	6	90	48.1	M	M	
DYS446	60	91	48.6	M	M	
DYS510	108	92	52.1	M	M	
DYS460	26	93	60.0	M	M	
DYS504	99	94	64.5	M	M	
Y-GATA-A10	89	95	66.2	M	M	
DYS570	33	96	76.7	F	F	
DYS714	73	97	85.3	M	F	
DYS532	97	98	87.4	M	F	
DYS715	98	99	88.2	M	F	
DYS534	55	100	88.8	F	F	
DYS458	13	101	89.6	F	F	
DYS650	96	102	94.6	F	F	
DYS549	78	103	99.6	M	F	
DYS442	36	104	100.3	M	M (from allele of 12) F (from allele of 13)	2
DYS449	21	105	103.0	F	F	
DYS533	82	106	107.4	M	M (from allele of 12) F (from allele of 13)	3
CDYa	34	107	148.0	VF	VF	
DYS576	32	108	156.2	VF	VF	
DYS710	68	109	208.3	VF	VF	
CDYb	35	110	208.7	VF	VF	
DYS712	94	111	252.2	VF	VF	

Note 1: The allele for DYS389-2 includes the count for 389-1

Note 2: The R-L21 Group A / Irish Type II tree shows a 12 to 13 mutation at DYS442 during the formative centuries of the Irish Type II haplotype (in the Bronze Age). The much higher frequency of mutations at DYS 442 seen only on this tree is due to subsequent back mutations to an allele of 12. It is placed 76th in the order of frequency of occurrence for the R-L21 Group B / Z16526 tree alone and the frequency categorisation as 'M' is consistent with observation elsewhere.

Note 3: A 12 to 13 mutation at DYS533 appears to have occurred between the time of SNP R-P312 and R-L21. Over 70% of mutations from an allele of 13 are back to 12. A lower frequency at this locus may be experienced elsewhere (e.g in R-U106).