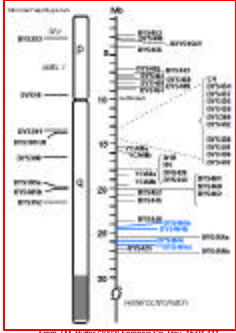


John M. Butler¹ and Richard Schoske^{1,2}

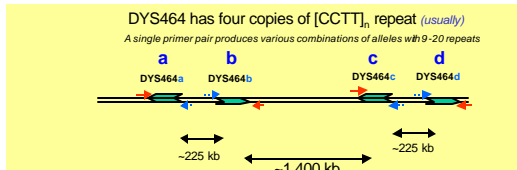
¹U.S. National Institute of Standards and Technology, Gaithersburg, MD; ²Current address: Armed Forces Institute of Pathology, Washington, DC

The Y-chromosome short tandem repeat (Y-STR) marker DYS464 was first reported by Redd *et al.* *Forensic Sci. Int.* (2002) 130:97-111 and appears to be the most polymorphic Y-STR marker discovered to date. A single primer pair can generate up to four distinct peaks. A careful mapping of DYS464 primers into the human genome reveals at least four copies occur over a 1.8 megabase (Mb) stretch near the DAZ region around 25 Mb on the Y-chromosome. Allele calls can be made based on peaks that are present (conservative approach) or a combination of alleles and peak height ratios (expanded typing method). However, the multitude of possible two and three peak patterns can potentially make this marker difficult to reliably type if mixtures from multiple males are involved. Issues of peak height ratio consistency will be examined in the context of different DYS464 patterns and discussed in terms of deciphering mixtures and potential degraded DNA profile patterns. We have observed 113 different DYS464 peak patterns using the conservative approach and 179 with the expanded typing method in 679 males from three U.S. populations. By comparison, in the same data set only 56 unique DYS385 types were seen. Perhaps more importantly, the addition of DYS464 results to the 9-locus minimal haplotype resulted in the ability to resolve almost all of the samples in our data set possessing the most common type that occurs in the European Y-STR Haplotype Reference Database (www.ystr.org). Several primer pairs have been developed for DYS464 and included in new multiplex assays.



Commonly used Y-STR markers have been mapped to chromosomal positions using the Human Genome Reference sequence assembly from April 2003 (<http://genome.ucsc.edu/cgi-bin/hgBlat>)

DYS464, a quadruplicated locus, occurs in the DAZ region near 25 Mb. This region was demonstrated to have palindromic sequences in the recent publication of the human Y-chromosome sequence (Skalesky *et al.*, (2003) *Nature* 423:825-837).



DYS464 has four copies of [CCTT]_n repeat (usually)
A single primer pair produces various combinations of alleles with 9-20 repeats

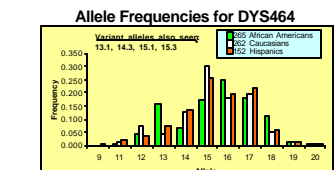
Expanded (E-type) 17,17,17,17 | 15,15,17,17 | 12,14,14,16 | 14,15,16,17
Conservative (C-type) 17 | 15,17 | 12,14,16 | 14,15,16,17

Example of some samples with same C-type that can be separated by E-type

C-type: 14,15,18

E-type

14,14,15,18
14,15,15,18
14,15,18,18



All DYS464 Expanded Types Observed with 679 Samples

Expanded Type	AA	EU	HP	Freq.	Expanded Type	AA	EU	HP	Freq.
14,15,16,16,16,16	1	1	1	0.003	14,14,14,14	1	1	1	0.001
14,15,16,17	2	1	1	0.003	14,14,14,15	1	1	1	0.001
14,15,16,18	2	1	1	0.003	14,14,14,16	1	1	1	0.001
14,15,16,19	1	1	1	0.001	14,14,14,17	1	1	1	0.001
14,15,16,20	1	1	1	0.001	14,14,14,18	1	1	1	0.001
14,15,17,17	3	1	1	0.003	14,14,14,19	1	1	1	0.001
14,15,17,18	3	1	1	0.003	14,14,14,20	1	1	1	0.001
14,15,17,19	2	1	1	0.001	14,14,15,15	1	1	1	0.001
14,15,17,20	1	1	1	0.001	14,14,15,16	1	1	1	0.001
14,15,18,18	2	1	1	0.003	14,14,15,17	1	1	1	0.001
14,15,18,19	1	1	1	0.001	14,14,15,18	1	1	1	0.001
14,15,18,20	1	1	1	0.001	14,14,15,19	1	1	1	0.001
14,15,19,19	1	1	1	0.001	14,14,15,20	1	1	1	0.001
14,15,20,20	1	1	1	0.001	14,14,16,16	1	1	1	0.001
14,16,16,16	1	1	1	0.001	14,14,16,17	1	1	1	0.001
14,16,16,18	1	1	1	0.001	14,14,16,19	1	1	1	0.001
14,16,16,20	1	1	1	0.001	14,14,17,17	1	1	1	0.001
14,16,17,17	1	1	1	0.001	14,14,17,18	1	1	1	0.001
14,16,17,19	1	1	1	0.001	14,14,17,20	1	1	1	0.001
14,16,17,20	1	1	1	0.001	14,14,18,18	1	1	1	0.001
14,16,18,18	1	1	1	0.001	14,14,18,19	1	1	1	0.001
14,16,18,20	1	1	1	0.001	14,14,18,20	1	1	1	0.001
14,16,19,19	1	1	1	0.001	14,14,19,19	1	1	1	0.001
14,16,20,20	1	1	1	0.001	14,15,15,15	1	1	1	0.001
14,17,17,17	1	1	1	0.001	14,15,15,16	1	1	1	0.001
14,17,17,18	1	1	1	0.001	14,15,15,17	1	1	1	0.001
14,17,17,19	1	1	1	0.001	14,15,15,18	1	1	1	0.001
14,17,17,20	1	1	1	0.001	14,15,15,19	1	1	1	0.001
14,17,18,18	1	1	1	0.001	14,15,15,20	1	1	1	0.001
14,17,18,19	1	1	1	0.001	14,15,16,16	1	1	1	0.001
14,17,18,20	1	1	1	0.001	14,15,16,17	1	1	1	0.001
14,17,19,19	1	1	1	0.001	14,15,16,18	1	1	1	0.001
14,17,20,20	1	1	1	0.001	14,15,16,19	1	1	1	0.001
14,18,18,18	1	1	1	0.001	14,15,16,20	1	1	1	0.001
14,18,19,19	1	1	1	0.001	14,15,17,17	1	1	1	0.001
14,18,20,20	1	1	1	0.001	14,15,17,18	1	1	1	0.001
14,19,19,19	1	1	1	0.001	14,15,17,19	1	1	1	0.001
14,19,20,20	1	1	1	0.001	14,15,17,20	1	1	1	0.001
14,20,20,20	1	1	1	0.001	14,15,18,18	1	1	1	0.001
14,20,21,21	1	1	1	0.001	14,15,18,19	1	1	1	0.001
14,20,22,22	1	1	1	0.001	14,15,18,20	1	1	1	0.001
14,21,21,21	1	1	1	0.001	14,15,19,19	1	1	1	0.001
14,21,22,22	1	1	1	0.001	14,15,19,20	1	1	1	0.001
14,22,22,22	1	1	1	0.001	14,15,20,20	1	1	1	0.001
14,23,23,23	1	1	1	0.001	14,16,16,16	1	1	1	0.001
14,24,24,24	1	1	1	0.001	14,16,16,17	1	1	1	0.001
14,25,25,25	1	1	1	0.001	14,16,16,18	1	1	1	0.001
14,26,26,26	1	1	1	0.001	14,16,16,19	1	1	1	0.001
14,27,27,27	1	1	1	0.001	14,16,16,20	1	1	1	0.001
14,28,28,28	1	1	1	0.001	14,16,17,17	1	1	1	0.001
14,29,29,29	1	1	1	0.001	14,16,17,18	1	1	1	0.001
14,30,30,30	1	1	1	0.001	14,16,17,19	1	1	1	0.001
14,31,31,31	1	1	1	0.001	14,16,17,20	1	1	1	0.001
14,32,32,32	1	1	1	0.001	14,16,18,18	1	1	1	0.001
14,33,33,33	1	1	1	0.001	14,16,18,19	1	1	1	0.001
14,34,34,34	1	1	1	0.001	14,16,18,20	1	1	1	0.001
14,35,35,35	1	1	1	0.001	14,16,19,19	1	1	1	0.001
14,36,36,36	1	1	1	0.001	14,16,19,20	1	1	1	0.001
14,37,37,37	1	1	1	0.001	14,16,20,20	1	1	1	0.001
14,38,38,38	1	1	1	0.001	14,17,17,17	1	1	1	0.001
14,39,39,39	1	1	1	0.001	14,17,17,18	1	1	1	0.001
14,40,40,40	1	1	1	0.001	14,17,17,19	1	1	1	0.001
14,41,41,41	1	1	1	0.001	14,17,17,20	1	1	1	0.001
14,42,42,42	1	1	1	0.001	14,17,18,18	1	1	1	0.001
14,43,43,43	1	1	1	0.001	14,17,18,19	1	1	1	0.001
14,44,44,44	1	1	1	0.001	14,17,18,20	1	1	1	0.001
14,45,45,45	1	1	1	0.001	14,17,19,19	1	1	1	0.001
14,46,46,46	1	1	1	0.001	14,17,19,20	1	1	1	0.001
14,47,47,47	1	1	1	0.001	14,17,20,20	1	1	1	0.001
14,48,48,48	1	1	1	0.001	14,18,18,18	1	1	1	0.001
14,49,49,49	1	1	1	0.001	14,18,18,19	1	1	1	0.001
14,50,50,50	1	1	1	0.001	14,18,18,20	1	1	1	0.001
14,51,51,51	1	1	1	0.001	14,18,19,19	1	1	1	0.001
14,52,52,52	1	1	1	0.001	14,18,19,20	1	1	1	0.001
14,53,53,53	1	1	1	0.001	14,18,20,20	1	1	1	0.001
14,54,54,54	1	1	1	0.001	14,19,19,19	1	1	1	0.001
14,55,55,55	1	1	1	0.001	14,19,19,20	1	1	1	0.001
14,56,56,56	1	1	1	0.001	14,20,20,20	1	1	1	0.001
14,57,57,57	1	1	1	0.001	14,21,21,21	1	1	1	0.001
14,58,58,58	1	1	1	0.001	14,22,22,22	1	1	1	0.001
14,59,59,59	1	1	1	0.001	14,23,23,23	1	1	1	0.001
14,60,60,60	1	1	1	0.001	14,24,24,24	1	1	1	0.001
14,61,61,61	1	1	1	0.001	14,25,25,25	1	1	1	0.001
14,62,62,62	1	1	1	0.001	14,26,26,26	1	1	1	0.001
14,63,63,63	1	1	1	0.001	14,27,27,27	1	1	1	0.001
14,64,64,64	1	1	1	0.001	14,28,28,28	1	1	1	0.001
14,65,65,65	1	1	1	0.001	14,29,29,29	1	1	1	0.001
14,66,66,66	1	1	1	0.001	14,30,30,30	1	1	1	0.001
14,67,67,67	1	1	1	0.001	14,31,31,31	1	1	1	0.001
14,68,68,68	1	1	1	0.001	14,32,32,32	1	1	1	0.001
14,69,69,69	1	1	1	0.001	14,33,33,33	1	1	1	0.001
14,70,70,70	1	1	1	0.001	14,34,34,34	1	1	1	0.001
14,71,71,71	1	1	1	0.001	14,35,35,35	1	1	1	0.001
14,72,72,72	1	1	1	0.001	14,36,36,36	1	1	1	0.001
14,73,73,73	1	1	1	0.001	14,37,37,37	1	1	1	0.001
14,74,74,74	1	1	1	0.001	14,38,38,38	1	1	1	0.001
14,75,75,75	1	1	1	0.001	14,39,39,39	1	1	1	0.001
14,76,76,76	1	1	1	0.001	14,40,40,40	1	1	1	0.001
14,77,77,77	1	1	1	0.001	14,41,41,41	1	1	1	0.001
14,78,78,78	1	1	1	0.001	14,42,42,42	1	1	1	0.001
14,79,79,79	1	1	1	0.001	14,43,43,43	1	1	1	0.001
14,80,80,80	1	1	1	0.001	14,44,44,44	1	1	1	0.001
14,81,81,81	1	1	1	0.001	14,45,45,45	1	1	1	0.001
14,82,82,82	1	1	1	0.001	14,46,46,46	1	1	1	0.001
14,83,83,83	1	1	1	0.001	14,47,47,47	1	1	1	0.001
14,84,84,84	1	1	1	0.001	14,48,48,48	1	1	1	0.001
14,85,85,85	1	1	1	0.001	14,49,49,49	1	1	1	0.001
14,86,86,86	1	1	1	0.001	14,50,50,50	1	1	1	0.001
14,87,87,87	1	1	1	0.001	14,51,51,51	1	1	1	0.001
14,88,88,88	1	1	1	0.001	14,52,52,52	1	1	1	0.001
14,89,89,89	1	1	1	0.001	14,53,53,53	1	1	1	0.001
14,90,90,90	1	1	1	0.001	14,54,54,54	1	1	1	0.001
14,91,91,91	1	1	1	0.001	14,55,55,55	1	1	1	0.001
14,92,92,92	1	1	1	0.001					