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Concordance Study Between the Investigator Argus Y-12 QS and the AmpFISTR® Yfiler™ Kits in a Brazilian Population Sample

Gustavo Chemale[1], Lídia dos Santos Rosa[2],
Jorge Marcelo de Freitas[3], Jeferson Loureiro Badaraco[4]

[1] *Doutor em Biologia Celular e Molecular*
[2] *Graduada em Ciências Biológicas*
[3] *Doutor em Ciências Biológicas*
[4] *Mestre em Ciências Biológicas*

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Abstract. Y-chromosome short tandem repeat (STR) analysis is an important tool in forensic casework, especially in sexual assault cases. As Y-STR genetic profiles might be shared and compared between different laboratories, states and even countries, concordance of allele calls among commercial kits is an important consideration. In this work, we investigated the concordance of allele calls in twelve Y-chromosome STR loci (DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439) between the Investigator Argus Y-12 QS (Qiagen) and the AmpFlSTR Yfiler™ (Life Technologies) kits in a sample of 155 male blood donors from Brasília, Federal District, Brazil. 155 samples previously typed with the Yfiler™ kit [Forensic Sci. Int. Genet. S1872-4973 (2013) 139-73] were also typed with the Investigator Argus Y-12 QS kit. A total of 152 different haplotypes were observed with the Yfiler™ kit, 149 being unique and three observed twice, while 145 different haplotypes were obtained with Argus Y-12, 135 being unique and ten observed twice in the population studied. A Different allele call was observed in only one sample at the DYS385 locus as 14-17 using the Yfiler™ kit and as 14-16.3 when using Argus Y-12. Haplotype diversity (HD) was calculated as 0.9992 for the results with the Investigator Argus Y-12 and as 0.9997 for the results with Yfiler™. Data obtained demonstrates that apart from the minor differences observed, both kits yielded concordant results for the Brazilian population analyzed.

Keywords: Y-STRs; Population data; Concordance study; Brazil; Haplotype diversity.

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1. Introdução

Y-chromosome short tandem repeat (STR) analysis is an important tool in forensic casework, especially in sexual assault cases. As Y-STR genetic profiles might be shared and compared between different laboratories, states and even countries, concordance of allele calls among different commercial kits is an important consideration.

We have previously typed 300 samples from male blood donors from Brasília, Federal District, Brazil with the AmpFlSTR Yfiler™ kit (Life Technologies) [1]. In this work, we investigated the concordance of allele calls in twelve Y-chromosome STR loci (DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439) between the Investigator Argus Y-12 QS kit (Qiagen) and Yfiler™ (Life Technologies) in 155 previously typed samples.

2. Methods

2.1 DNA extraction

Genomic DNA was extracted from blood samples using the salting out protocol [2] and quantified for human male DNA using the Quantifiler™ Y Human Male DNA Quantification Kit (Life Technologies) on an iQ5 Multicolor Real-Time PCR Detection System (Bio-Rad) as previously published [1].

2.2 Y-STR amplification

Y-Chromosome STRs were amplified from approximately 1 ng of template DNA. Samples were previously typed using the AmpFlSTR® Yfiler™ PCR Amplification Kit (Life Technologies) [1] and in this work typed with the Investigator Argus Y-12 QS kit, according to the manufacturer's instructions.

2.3 STR typing

STR typing was performed in an ABI 3130 Genetic Analyzer (Life Technologies) using GeneMapper ID-X v. 1.1.2 analysis software. All electropherograms were also individually checked for quality and allele call confirmation. Samples showing biallelic, triallelic or null allele patterns were re-amplified for confirmation.

2.4 Statistics

Haplotype frequencies were estimated by the counting method. Haplotype diversity was calculated using ARLEQUIN version 3.5.1.3 software [3], considering twelve loci

for the results with the Investigator Argus Y-12 QS kit and seventeen loci for the Yfiler™ kit.

3. Results and discussion

A total of 155 samples previously typed with the Yfiler™ kit [1] were also typed with the Investigator Argus Y-12 QS kit. 152 different haplotypes were observed with the Yfiler™ kit, 149 being unique and three observed twice (Table 1), while 145 different haplotypes were obtained with Argus Y-12, 135 being unique and ten observed twice in the population studied (Table 2). Haplotype diversity (HD) was calculated as 0.9992 for the results with the Investigator Argus Y-12 and as 0.9997 for the results with Yfiler™.

One different allele call was observed in only one loci when results for both kits were compared. One sample was typed at the DYS385 locus as 14-17 using the Yfiler™ kit and as 14-16.3 when using Argus Y-12 (Figure 1). This difference has been reported previously [4] and is also mentioned in the kit´s manual that allele 16.3 represents allele 17 with one thymidine deletion between the primer binding site and the repeat region [5]. This deletion is not within the region amplified using the primers in the Yfiler™ kit [6].

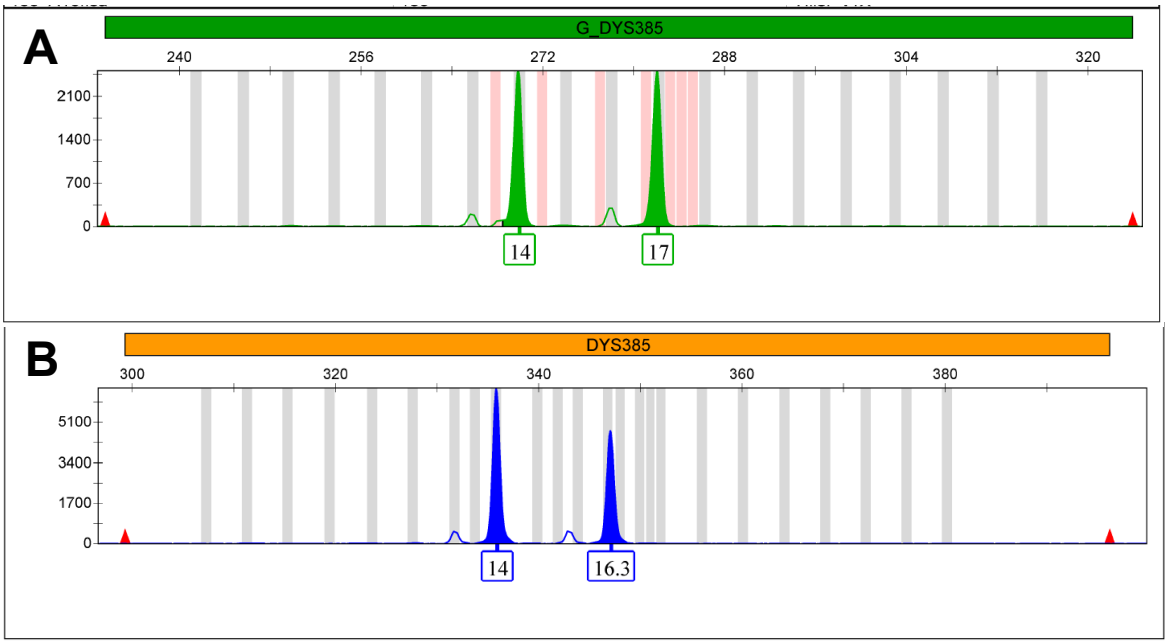

Figure 1. Allelic microvariant called in DYS385 locus with different kits. **A.** Argus Y-12 (Qiagen) and **B.** Yfiler (Life Technologies).

Haplotypes with double peaks at DYS391 and DYS439, besides those at the DYS385 locus were observed with both kits. Moreover, a tri-allelic pattern and a null allele were also observed at the DYS385 locus with both kits.

The data obtained demonstrates that apart from the minor differences observed, both kits yielded concordant results for the Brazilian population analyzed. Analysts should take these differences into account when comparing profiles obtained with these kits or in case of databasing. The Investigator Argus Y-12 QS was able to detect less different haplotypes and showed a high but slightly lower haplotype diversity than Y-filer™.

Population dataset was previously published [1] and is available in the Y-STR Haplotype reference database (http://www.yhrd.org) under the accession number YA003751.

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Table 1. Y-chromosome haplotypes obtained with Yfiler in 155 blood donors from Brasília. Data shown is limited to the markers shared with Argus Y-12. Haplotype frequency was calculated taking into consideration the seventeen loci type with the Yfiler kit.

Haplotype	N	DYS439	DYS437	DYS390	DYS385a/b	DYS391	DYS389I	DYS19	DYS389II	DYS393	DYS438	DYS392
1	1	12	14	22	16,17	10	14	16	31	14	11	11
2	1	11	15	24	11,14	12	13	14	29	13	12	13
3	1	13	14	23	11,14	11	13	14	29	13	12	13
4	1	12	15	24	11	11	13	14	29	13	12	13
5	1	11	15	23	11,14	11	13	14	29	13	12	13
6	1	12	14	23	14	10	14	14	31	12	9	12
7	1	12	16	22	15	10	12	15	30	13	10	11
8	1	13	15	22	15	10	12	16	29	14	10	11
9	1	11	15	23	15	11	13	16	29	14	10	12
10	2	12	15	24	11,13	11	14	14	30	13	12	12
11	1	12	15	23	11,15	11	13	15	29	13	12	13
12	1	11	15	25	12,16	10	13	15	29	13	10	11
13	1	12	14	21	17,18	10	13	17	30	14	11	11
14	1	12	14	23	16,17	10	12	15	29	13	10	11
15	1	11	14	21	16,17	11	13	15	31	13	11	11
16	1	13	15	24	11,15	11	13	14	29	13	12	13
17	1	13	15	22	13,15,17	10	14	13	31	13	9	13
18	1	11	14	21	16,17	11	13	16	31	13	11	11
19	1	12	14	24	13,16	9	13	15	29	12	9	11
20	1	12	15	24	11,14	11	14	14	30	13	12	13
21	1	12	14	24	16,19	10	13	13	29	13	10	11
22	1	13	14	24	11,14	11	13	14	29	13	12	13
23	1	13	15	23	11,15	11	13	14	29	13	12	13
24	1	12	14	21	17,19	10	15	14	34	12	11	10
25	1	12	14	24	16	10	14	13	33	12	10	11
23	1	14	15	24	12.2,14	11	14	14	29	12	12	14
27	1	12	15	24	11,16	10	13	14	29	13	12	13
28	1	12	14	24	16,17	10	13	13	30	13	10	11

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29	1	12	14	25	11,14	11	13	14	29	13	12	13
30	1	11	14	23	11,13	11	13	14	29	13	12	13
31	1	13	15	25	11,14	10	15	14	31	13	12	14
32	1	12	15	24	11,14	11	13	14	29	13	12	13
33	1	12	14	24	16,18	10	13	13	30	12	9	11
34	1	12	16	21	15,16	10	12	15	28	16	10	11
35	1	12	16	22	13,16	11	12	15	28	12	9	11
36	1	10	14	24	14,18	10	12	13	30	13	11	14
37	1	11	14	24	14,18	10	12	13	30	13	11	14
38	1	13	15	24	11,12	11	13	14	29	12	13	13
39	1	11	15	24	11,14	11	13	14	29	13	12	13
40	1	12	14	22	14,15	10	13	13	29	13	12	14
41	1	11	14	23	16,19	10	13	13	31	13	10	11
42	1	12	16	22	12,15	10	12	16	28	13	10	11
43	1	12	14	24	12,14	11	13	14	31	13	12	13
44	1	13	14	24	16	10	13	13	30	12	10	11
45	1	12	15	24	11,14	11	13	14	29	12	12	13
46	2	12	14	24	11,14	11	13	14	29	13	12	13
47	1	12	15	24	12,15	11	13	14	29	13	12	13
48	2	12	14	22	13,16	9	13	15	29	12	9	11
49	1	13	15	24	12,14	11	13	14	30	13	12	13
50	1	13	15	25	12,13	11	14	14	31	13	12	13
51	1	11	15	24	13,15	10	14	15	29	13	11	13
52	1	12	14	23	13,17	9	13	15	29	12	9	11
53	1	12	15	24	11,14	11	10	14	26	13	12	14
54	1	11	14	21	17,19	10	15	16	32	15	11	11
55	1	11	15	24	11,14	10	12	15	29	13	12	13
56	1	11	14	21	16,17	11	13	15	30	13	11	11
57	1	11	16	23	13,14	11	12	14	28	13	10	11
58	1	12	15	23	13,18	10	13	15	29	12	9	11
59	1	11	15	22	13,15	10	13	14	30	12	9	11

60	1	11	14	23	14,15	10	14	15	31	14	10	12
61	1	13	14	24	14,15	11	12	14	29	13	11	14
62	1	11	14	24	15,18	10	13	13	29	13	11	14
63	1	11,13	14	22	12,16	9	13	14	30	13	10	11
64	1	12	15	24	10,15	10	13	14	29	13	12	13
65	1	10	14	23	13,14	9	13	14	30	13	10	11
66	1	10	14	23	13,14	9	13	14	30	13	10	11
67	1	13	15	24	11,15	11	13	14	29	14	12	13
68	1	13	14	21	16,18	10	14	16	31	15	11	11
69	1	11	15	23	11,15	11	13	14	29	13	12	13
70	1	11	14	23	13,18	11	13	14	30	12	10	11
71	1	11	16	23	13,15	10	12	14	28	13	10	11
72	1	11	16	22	14	11	12	15	29	13	10	11
73	1	11	15	24	11,14	11	12	14	29	13	12	13
74	1	11,13	14	22	12,15	9	13	13	30	13	10	11
75	1	12	15	25	11,14	11	12	15	28	13	12	13
76	1	11	16	21	13,17	10	12	15	29	15	10	11
77	1	12	15	24	11,13	11	13	14	29	13	12	13
78	1	11	15	23	15	11	13	16	29	14	10	12
79	1	12	15	24	12,14	11	13	14	29	13	12	13
80	1	12	14	23	13,16	9	13	15	29	12	9	11
81	1	12	14	23	14	10	14	15	31	12	9	13
82	1	11	14	21	17,18	11	13	15	30	13	11	11
83	1	11	14	23	13,18	10	13	14	29	12	10	11
84	1	13	15	24	11,14	10	13	14	29	13	12	13
85	1	11	16	23	14	10	12	15	30	14	9	11.3
86	1	12	15	24	12,13	11	13	14	29	13	12	13
87	1	12	13	21	16,17	10	13	16	30	15	11	11
88	1	12	14	24	16,18	10	12	13	29	13	10	11
89	1	12	15	23	11,14	11	13	14	29	14	12	13
90	1	11	15	24	11,15	12	13	14	29	13	12	13

91	1	11	15	23	11,14	11	13	14	29	13	12	13
92	1	11	15	25	11	10	13	14	28	13	12	13
93	1	12	15	24	11,14	10	12	14	27	13	12	13
94	1	12	15	23	13,20	10	13	15	29	12	9	11
95	1	13	15	24	11,14	10	13	15	29	14	12	14
96	1	12	14	24	16,17	10	12	15	29	13	10	11
97	1	11	15	23	12	10	13	17	28	13	10	11
98	1	11	14	21	16,17	11	13	15	31	13	11	11
99	1	12	14	23	16,17	10	13	15	30	13	10	11
100	1	12	14	24	15,17	10	13	13	30	13	10	11
101	1	11	15	24	11,14	11	14	14	30	13	12	13
102	1	12	14	23	11,14	11	15	14	31	13	12	13
103	1	12	15	24	12,13	11	13	13	29	13	12	13
104	1	13	15	23	13,17	10	13	14	29	12	9	11
105	1	12	15	24	11,14	11	12	14	28	13	12	13
106	1	10	14	23	11,13	11	13	15	29	14	10	14
107	1	12	15	25	11,14	11	13	14	29	13	12	13
108	1	11	16	24	13,15	10	12	15	28	13	10	11
109	1	12	15	22	16,17	10	14	16	31	14	10	12
110	1	13	15	24	12.2,14	11	14	14	29	11	12	14
111	1	10	14	24	11,14	10	14	16	31	14	11	11
112	1	13	15	24	11,13	11	13	14	29	13	12	14
113	1	11	16	23	16,17	10	14	14	30	12	10	14
114	1	12	14	21	17,18	9	13	17	30	15	11	11
115	1	13	15	24	11,14	11	13	14	29	13	12	13
116	1	10	14	23	13,14	9	14	14	31	12	10	11
117	1	13	15	24	11,14	11	13	14	29	12	12	14
118	1	13	15	24	11,14	11	13	15	29	13	10	13
119	1	12	15	24	11,14	11	13	14	29	13	12	13
120	1	12	15	24	11,15	11	13	14	29	13	12	13
121	1	12	14	25	11,14	11	13	14	29	13	12	13

122	1	12	14	25	11,14	11	12	14	29	13	12	13
123	1	11	14	25	11,14	11	13	14	30	13	12	13
124	1	11	14	24	16,18	10	13	13	30	13	10	11
125	1	11	14	24	12,14	10	13	14	29	14	12	13
126	1	11	14	23	15,18	10	13	17	30	13	9	14
127	1	11	14	25	14,19	11	12	14	28	13	12	11
128	1	10	14	23	11,14	11	13	15	29	14	10	14
129	1	13	14	23	14,17	11	12	13	29	13	11	15
130	1	10	14	23	13,14	9	14	14	31	13	10	11
131	1	13	16	22	13	10	14	15	32	13	10	11
132	1	11	14	25	11,14	11	13	16	30	13	11	11
133	1	11	15	25	11,14	11	14	14	31	13	12	13
134	1	11	16	22	13,16	11	12	15	29	12	9	11
135	1	12	15	23	11,14	11	13	14	29	13	12	13
136	1	10	15	23	14	10	12	16	29	15	10	12
137	1	10	15	24	12,14	11	12	16	26	13	12	13
138	1	12	14	24	13,14	10	14	14	30	13	11	14
139	1	10	14	24	13,14	9	14	13	30	13	10	11
140	1	11	15	23	13,15	10	13	15	30	15	10	12
141	1	12	15	23	11,14	11	13	14	29	13	12	13
142	1	11	15	24	11,14	11	12	15	28	13	12	13
143	1	10	14	24	13,14	9	12	13	28	13	10	11
144	1	11	16	24	14,17	9	12	15	28	13	9	11
145	1	11	16	24	14,15	10	12	15	28	13	10	11
146	1	12	14	24	16,18	9	14	13	31	14	10	11
147	1	11	13	23	14	10	13	13	29	13	11	15
148	1	12	15	23	11,14	11	13	16	29	13	12	13
149	1	11	14	24	11,13	10	14	14	30	14	12	13
150	1	11,12	14	23	12,17	9,10	13	13	30	13	10	11
151	1	11	14	24	null	11	13	14	31	13	12	13
152	1	14	15	24	11	10	14	15	33	13	10	11

Table 2. Y-chromosome haplotypes obtained with Argus Y-12 in 155 blood donors from Brasília.

Haplotype	N	DYS439	DYS437	DYS390	DYS385	DYS391	DYS389I	DYS19	DYS389II	DYS393	DYS438	DYS392
1	1	12	14	22	16,17	10	14	16	31	14	11	11
2	1	11	15	24	11,14	12	13	14	29	13	12	13
3	1	13	14	23	11,14	11	13	14	29	13	12	13
4	1	12	15	24	11	11	13	14	29	13	12	13
5	2	11	15	23	11,14	11	13	14	29	13	12	13
6	1	12	14	23	14	10	14	14	31	12	9	12
7	1	12	16	22	15	10	12	15	30	13	10	11
8	1	13	15	22	15	10	12	16	29	14	10	11
9	2	11	15	23	15	11	13	16	29	14	10	12
10	2	12	15	24	11,13	11	14	14	30	13	12	12
11	1	12	15	23	11,15	11	13	15	29	13	12	13
12	1	11	15	25	12,16	10	13	15	29	13	10	11
13	1	12	14	21	17,18	10	13	17	30	14	11	11
14	1	12	14	23	16,17	10	12	15	29	13	10	11
15	2	11	14	21	16,17	11	13	15	31	13	11	11
16	1	13	15	24	11,15	11	13	14	29	13	12	13
17	1	13	15	22	13,15,17	10	14	13	31	13	9	13
18	1	11	14	21	16,17	11	13	16	31	13	11	11
19	1	12	14	24	13,16	9	13	15	29	12	9	11
20	1	12	15	24	11,14	11	14	14	30	13	12	13
21	1	12	14	24	16,19	10	13	13	29	13	10	11
22	1	13	14	24	11,14	11	13	14	29	13	12	13
23	1	13	15	23	11,15	11	13	14	29	13	12	13
24	1	12	14	21	17,19	10	15	14	34	12	11	10
25	1	12	14	24	16	10	14	13	33	12	10	11
26	1	14	15	24	12.2,14	11	14	14	29	12	12	14
27	1	12	15	24	11,16	10	13	14	29	13	12	13

28	1	12	14	24	16,17	10	13	13	30	13	10	11
29	2	12	14	25	11,14	11	13	14	29	13	12	13
30	1	11	14	23	11,13	11	13	14	29	13	12	13
31	1	13	15	25	11,14	10	15	14	31	13	12	14
32	2	12	15	24	11,14	11	13	14	29	13	12	13
33	1	12	14	24	16,18	10	13	13	30	12	9	11
34	1	12	16	21	15,16	10	12	15	28	16	10	11
35	1	12	16	22	13,16	11	12	15	28	12	9	11
36	1	10	14	24	14,18	10	12	13	30	13	11	14
37	1	11	14	24	14,18	10	12	13	30	13	11	14
38	1	13	15	24	11,12	11	13	14	29	12	13	13
39	1	11	15	24	11,14	11	13	14	29	13	12	13
40	1	12	14	22	14,15	10	13	13	29	13	12	14
41	1	11	14	23	16,19	10	13	13	31	13	10	11
42	1	12	16	22	12,15	10	12	16	28	13	10	11
43	1	12	14	24	12,14	11	13	14	31	13	12	13
44	1	13	14	24	16	10	13	13	30	12	10	11
45	1	12	15	24	11,14	11	13	14	29	12	12	13
46	2	12	14	24	11,14	11	13	14	29	13	12	13
47	1	12	15	24	12,15	11	13	14	29	13	12	13
48	2	12	14	22	13,16	9	13	15	29	12	9	11
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