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## Announcement of Population Data

## Y-STR variation among ethnic groups from Ecuador: Mestizos, Kichwas, Afro-Ecuadorians and Waoranis

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## ABSTRACT

Twelve Y-chromosomal short tandem repeats (STRs) DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385ab, DYS437, DYS438 and DYS439 were studied in the three major ethnic groups from Ecuador: Mestizos, Native Amerindians (Kichwas, Quichuas) and Afro-Ecuadorians aiming to construct a representative database for this region in Latin America. All three populations exhibit high haplotypes diversities. Analysis of molecular variance (AMOVA) reveals significant differentiation between the Mestizos, the Kichwas and the Afro-Ecuadorians. The analysis of a hunter-gatherer group of Native Amerindians from the Amazonian provinces of Ecuador, the Waoranis (Huaorani) revealed markedly reduced haplotypes variability and a large genetic distance to the major Ecuadorian populations.

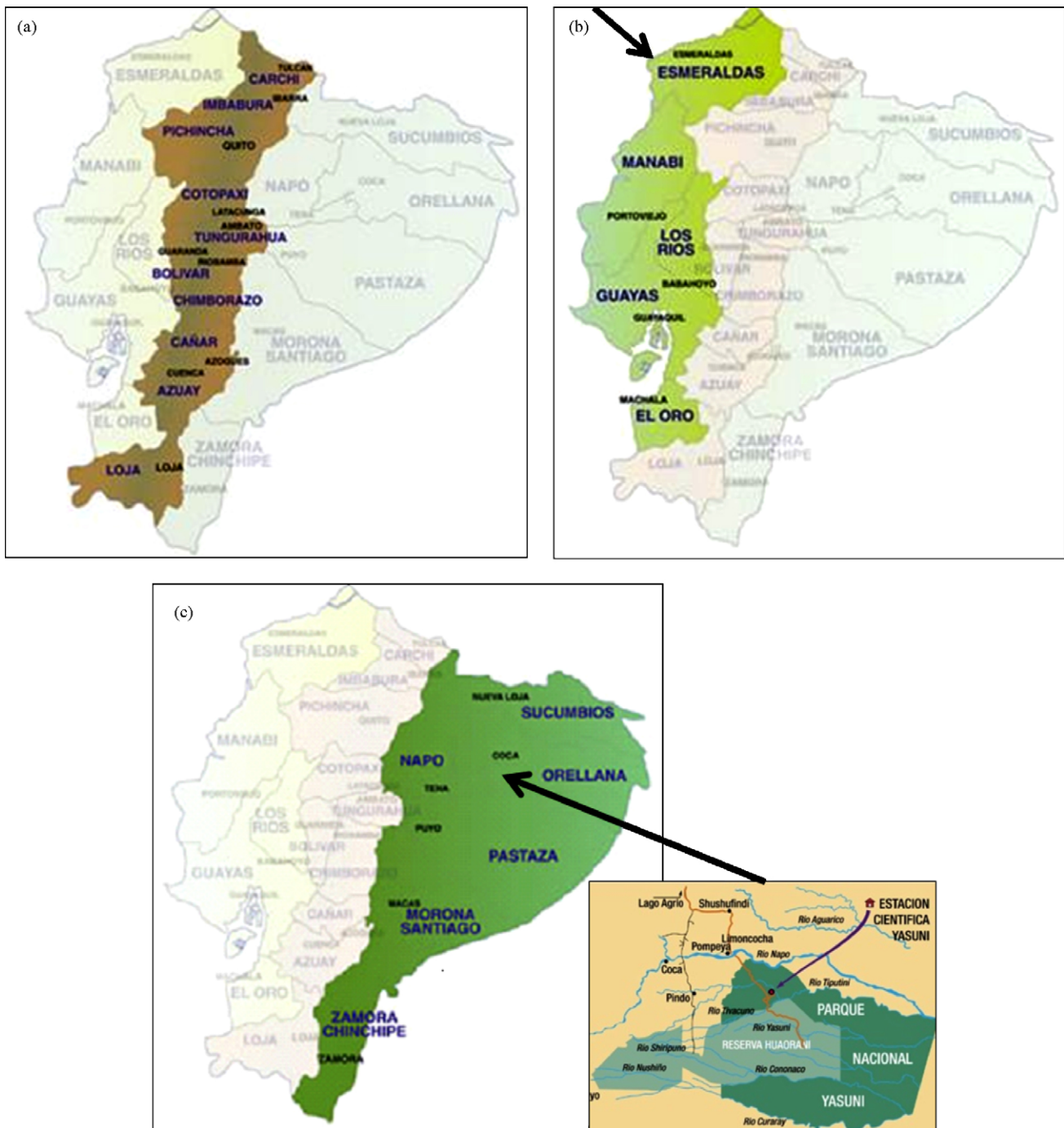
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## 1. Population

Whole blood was obtained in EDTA vacutainers tubes by venipuncture from 102 Mestizo, 102 Kichwa, 102 Afro-Ecuadorian and 35 Waorani males born and living in Ecuador. Samples from Kichwas, Afro-Ecuadorian and Waoranis were obtained directly at their communities. Samples of Mestizos were taken from a paternity test bank of our laboratory in Quito. All the samples were obtained after informed written consent and were approved by the Bioethics Committee of our hospital. We selected the individuals using criteria such as skin color, surnames, town of origin and language. All samples have been previously typed for autosomal short tandem repeats (STRs) [1–4]. Ecuador's population is ethnically diverse. The largest ethnic group is comprised of Mestizos, the mixed descendants of Spanish colonists and indigenous Amerindians, who constitute just over 65% of the population [5]. Native Amerindians (Kichwa speaking) are second in numbers and account for approximately a fifth of the current population, around 3 million people [6]. Most of them live in the mountains, in the Andean region, from Carchi province in the north to Loja province in the South

(Fig. 1a). Some of them, speaking a different Kichwa language, inhabit the Amazonian provinces of Pastaza, Napo, Sucumbios and Orellana. The small Afro-Ecuadorian minority (~500,000 individuals), is largely based in coastal Esmeraldas province and in the Valle de Chota located in the Andean region (Fig. 1b). The Waorani are the last nomad population of hunters and gatherers in Ecuador. Known traditionally as "Aucas" or "Jíbaros", they have been popularized as being capable of "shrinking heads", a process known as "tzantza". They speak the "Wao Tiro" language, which, according to linguistic studies, does not have any related languages. The Waoranis currently live in small isolated groups in Ecuadorian Amazonia, some of them subject to oil companies that are deforesting and destroying their natural environment, whilst other individuals live isolated without any contact with the Western world, for example the "Tagaeris", the "Taromenanes" and the "Oñamenanes". According to the Waorani Nationality Diagnosis carried out in 1999, the population exceeds 2200 people. They can be found in three provinces of Amazonia (Orellana, Pastaza and Napo, see Fig. 1(c)). Their traditional territory extended over an approximate area of 2,000,000 ha, between the right bank of the Napo River and the left bank of the Curaray. They are currently organized in around 28 communities which are grouped together around the population in contact [6]. This population maintained its independence and defend their territory through belligerent actions. The

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**Fig. 1.** Maps of Ecuador and location of the studied populations. Mestizos live predominantly in urban regions, whereas the Kichwa live in the central highlands (a), the Afro-Ecuadorians in the coastal Esmeraldas province (b) and the Waorani in the Eastern provinces of Napo, Pastaza and Orellana (c). Sampling location for the latter are marked with arrows.

traditional basic unit or domestic group is the “Nanicabo”, made up of extended or multiple families comprising between six and ten families which live under a same roof or “maloca”; these domestic groups are self-sufficient and autonomous, and are organized around an elderly member whose name derives from the name of the domestic group. The territorial unit or local group is known as “Waomoni”, which is the coming together of several “Nanicabos”. The “Waomonis” are of endogamic character, in which marriage is between crossed cousins and relations are of alliance. Large parts of the communities are formed by two or three “Waomonis” of diverse origin. A minority lives either in grouped together “malocas” or in

isolated domestic groups. Their ruling body is the *Bile Waorani Council*, which is the Assembly for all the nationality. The leading Organization for external representation is the Waorani Nationality of Ecuadorian Amazonia Organization (ONHAE) which has been in existence since the 1990s.

**2. Extraction**

Extraction was done using Wizard™ system (Promega Corporation, Madison, USA) and the quantity was estimated by UV absorbance.

**Table 1**  
Y chromosome STR haplotypes from Ecuador

Ht	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385a	DYS 437	DYS 438	DYS 439	Ecuador [Kichwa]	Ecuador [Mestizo]	Ecuador [Afro-Ecuadorian]	Ecuador [Waorani]
1	12	13	29	25	9	13	14	15.15	14	11	11	1			
2	12	13	30	23	10	14	13	15.17	13	10	12	1	2		
3	12	13	30	24	10	14	14	12.14	14	11	12	1			
4	13	12	28	24	10	14	13	14.16	14	11	12	1			
5	13	12	28	24	10	14	14	15.15	14	9	11	1			
6	13	12	29	23	9	14	14	14.18	14	11	11	1			
7	13	12	29	23	9	15	13	15.16	14	12	11	1			
8	13	12	29	23	10	15	13	15.16	14	12	12			1	
9	13	12	29	24	10	11	13	17.18	14	10	10		1		
10	13	12	29	24	10	14	13	13.18	13	11	13			1	
11	13	12	29	25	10	11	13	17.18	14	10	11		1		
12	13	12	29	25	10	14	13	14.17	14	11	12			1	
13	13	12	30	24	10	11	14	15.18	15	10	11		1		
14	13	12	30	24	10	14	13	14.18	14	11	13		1		
15	13	12	30	24	10	14	14	12.17	14	11	13	1			
16	13	12	30	24	10	14	14	12.17	14	12	12		1		
17	13	12	30	24	11	14	14	16.18	14	11	12	1			
18	13	12	30	25	10	10	13	18.19	14	9	10		1		
19	13	12	30	25	10	14	13	14.17	14	11	11	1			
20	<b>13</b>	<b>12</b>	<b>30</b>	<b>25</b>	<b>10</b>	<b>14</b>	<b>14</b>	<b>14.19</b>	<b>11</b>	<b>12</b>	<b>15</b>				<b>2</b>
21	13	12	31	24	10	13	13	16.18	14	11	13	1			
22	13	12	31	24	10	14	13	13.19	14	12	12			1	
23	<b>13</b>	12	31	24	10	15	13	15.16	14	12	11	1			
24	<b>13</b>	<b>13</b>	<b>28</b>	<b>23</b>	<b>10</b>	<b>14</b>	<b>13</b>	<b>15.15</b>	<b>11</b>	<b>10</b>	<b>14</b>				<b>9</b>
25	13	13	29	23	10	14	13	15.15	15	11	11	1	1		
26	13	13	29	23	10	14	15	14.17	14	9	12	1	1		
27	13	13	29	24	10	13	12	14.15	14	11	12				
28	13	13	29	24	10	13	13	14.14	14	11	12	1			
29	13	13	29	24	10	14	13	14.15	14	11	12	1			
30	13	13	29	24	10	14	13	15.16	14	11	12	2			
31	13	13	29	24	10	14	14	14.17	15	11	12	1	1		
32	13	13	29	24	10	15	13	15.16	14	11	12	1	1		
33	13	13	29	24	11	12	13	12.17	14	11	12		1		
34	13	13	29	24	11	13	13	13.21	14	11	12				
35	13	13	29	25	10	14	15	14.16	14	11	12	1			
36	13	13	29	25	11	14	14	14.15	14	11	12	1	1		
37	13	13	30	23	10	11	13	16.17	14	10	12	1			
38	13	13	30	23	10	14	13	15.15	14	11	12	1		1	
39	13	13	30	23	10	14	14	12.16	14	11	11		1		
40	13	13	30	23	10	14	14	14.14	14	10	12		1		
41	13	13	30	23	10	15	13	13.17	14	11	9				
42	13	13	30	23	10	15	13	15.17	14	11	12	1			
43	13	13	30	23	11	14	13	14.16	14	11	12	1		1	
44	13	13	30	23	11	14	13	14.17	14	11	12			1	
45	13	13	30	24	9	11	13	13.14	14	10	10				
46	13	13	30	24	9	13	13	14.14	14	11	12	1		1	
47	13	13	30	24	10	11	13	16.17	14	10	11			1	
48	13	13	30	24	10	11	13	16.18	14	10	12		1		
49	13	13	30	24	10	11	14	13.17	14	10	13				
50	13	13	30	24	10	12	13	13.17	14	11	12	1			
51	13	13	30	24	10	13	13	14.14	14	11	12	3			
52	13	13	30	24	10	14	11	15.17	14	11	12		1		
53	13	13	30	24	10	14	13	13.20	14	11	11				
54	13	13	30	24	10	14	13	14.18	14	11	11	1			
55	13	13	30	24	10	14	13	15.15	14	11	12	1			

Table 1 (Continued)

Ht	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385a	DYS 437	DYS 438	DYS 439	Ecuador [Kichwa]	Ecuador [Mestizo]	Ecuador [Afro-Ecuadorian]	Ecuador [Waorani]
56	13	13	30	24	10	14	13	16.16	14	11	12	1		1	
57	13	13	30	24	10	14	14	12.16	14	11	11				
58	13	13	30	24	10	15	13	14.15	14	11	13	1			
59	13	13	30	24	10	15	14	14.18	14	12	14	1			
60	13	13	30	24	10	17	13	14.14	14	11	12	1	1		
61	13	13	30	24	11	13	13	12.14	15	12	11				
62	13	13	30	24	11	14	13	12.16	14	11	12	1	1		
63	13	13	30	24	11	14	13	12.20	14	10	12				
64	13	13	30	24	11	14	13	13.20	14	11	11	1		1	
65	13	13	30	24	11	14	15	15.16	14	8	11		1		
66	13	13	30	25	10	14	13	15.16	14	11	12				
67	13	13	30	25	10	14	15	15.18	14	8	11	1			
68	13	13	31	23	10	11	13	15.17	14	10	12	1			
69	13	13	31	23	10	14	14	12.17	14	11	12	1			
70	13	13	31	24	9	13	13	14.18	14	11	13	1	1		
71	13	13	31	24	10	11	13	15.19	14	10	13				
72	13	13	31	24	10	13	12	13.15	14	11	13	1			
73	13	13	31	24	10	14	11	14.17	14	11	12	1	1		
74	13	13	31	24	10	14	14	12.17	14	11	13				
75	13	13	31	24	10	14	14	14.15	14	11	11	1	1		
76	13	13	31	24	11	11	13	16.17	14	10	13				
77	13	13	31	24	11	14	14	12.17	14	11	13	1			
78	13	13	31	25	10	14	14	14.18	14	11	12	1			
79	13	13	32	22	9	11	13	13.15	14	10	12	1	1		
80	13	13	32	24	11	14	14	12.17	14	11	12				
81	13	13	32	24	11	15	14	12.17	14	11	11	1			
82	13	13	32	25	9	14	14	12.19	14	11	12	1	1		
83	13	13	32	25	10	13	13	14.17	15	12	12			1	
84	13	14	30	24	9	11	13	14.16	14	10	10				
85	13	14	30	24	10	14	13	14.16	14	11	11	1			
86	13	14	30	24	10	15	13	11.18	14	11	12	1	1		
87	13	14	30	25	10	14	13	14.19	13	11	12				
88	13	14	30	25	11	14	13	13.19	14	11	12	1	1		
89	13	14	31	23	10	14	13	13.17	14	11	12				
90	13	14	31	23	10	14	13	13.17	14	11	13	1	1		
91	13	14	31	23	10	14	13	13.18	14	9	12				
92	13	14	31	23	10	14	13	15.18	14	11	11	1			
93	13	14	31	23	10	16	14	14.18	14	11	13	1			
94	13	14	31	23	10	16	14	15.19	14	11	13	1		1	
95	13	14	31	23	11	15	13	13.16	14	12	13			1	
96	13	14	31	24	10	11	13	16.18	14	10	11			1	
97	13	14	31	24	10	13	13	11.14	15	12	12		1		
98	13	14	31	24	10	14	13	13.18	14	11	12		1		
99	13	14	31	24	10	14	13	13.19	14	11	12				
100	13	14	31	24	10	14	13	14.19	14	11	12	1			
101	13	14	31	24	10	14	13	14.20	14	11	12	1	1		
102	13	14	31	24	10	14	14	15.16	14	11	13				
103	13	14	31	24	11	14	13	13.19	14	11	12	1		1	
104	13	14	31	24	11	14	13	14.19	14	11	11				
105	13	14	31	24	11	14	13	19.19	14	11	12	2	1		
106	13	14	31	24	11	14	14	13.17	14	11	11				
107	13	14	31	24	11	15	13	12.17	14	11	13	1		1	
108	13	14	31	25	10	14	12	14.15	15	11	11				
109	13	14	32	23	11	14	14	14.16	14	11	9	1			
110	13	14	32	24	11	14	13	13.19	15	11	13	1	1		
111	13	15	32	24	10	11	13	16.16	14	10	11		1		

112	14	12	28	22	10	11	13	13.15	16	10	11								
113	14	12	28	22	10	11	13	14.14	16	10	11	1							
114	14	12	28	22	11	14	13	15.17	15	11	12	1						1	
115	14	12	28	24	11	11	13	15.20	14	11	12			1					
116	14	12	28	24	11	13	13	11.14	15	12	15			1					1
117	14	12	28	25	11	11	13	14.17	15	11	11								1
118	14	12	28	25	11	13	13	11.15	15	12	12								
119	14	12	29	24	11	13	13	12.14	15	12	11	2							
120	14	12	30	23	10	15	12	14.16	12	12	14								9
121	14	12	31	24	10	15	13	15.15	14	12	12	1		1					
122	14	13	28	24	11	13	13	11.14	15	12	11			1					
123	14	13	28	24	12	13	13	11.14	15	12	12								1
124	14	13	28	24	13	13	13	11.14	15	12	12			1					1
125	14	13	29	22	10	13	13	12.14	15	12	11								1
126	14	13	29	22	11	13	12	11.14	15	12	13			1					
127	14	13	29	23	10	11	12	14.17	14	10	12								1
128	14	13	29	23	10	13	13	11.18	15	12	13			1					
129	14	13	29	24	10	12	13	11.14	15	12	13			1					
130	14	13	29	24	10	13	13	11.13	15	12	11			1					
131	14	13	29	24	10	13	13	12.13	15	12	11			1					
132	14	13	29	24	10	13	13	12.14	14	12	11			1					
133	14	13	29	24	10	13	13	12.14	14	12	12			1					
134	14	13	29	24	10	13	13	12.14	15	12	12								
135	14	13	29	24	10	14	13	16.19	14	11	11	1		1					
136	14	13	29	24	11	13	13	11.13	14	12	12			1					
137	14	13	29	24	11	13	13	11.14	14	12	11			1					
138	14	13	29	24	11	13	13	11.14	14	12	12	1		1					
139	14	13	29	24	11	13	13	11.14	15	10	13			1					
140	14	13	29	24	11	13	13	11.14	15	12	11			1					
141	14	13	29	24	11	13	13	11.14	15	12	12								
142	14	13	29	24	11	13	13	11.14	15	12	13	1		1					
143	14	13	29	24	11	13	13	11.14	15	12	14			1					
144	14	13	29	24	11	13	13	11.14	15	18	12			1					
145	14	13	29	24	11	13	13	11.15	15	12	13			1					
146	14	13	29	24	11	13	15	11.14	14	12	13			2					
147	14	13	29	24	11	14	13	11.14	14	12	12			1					
148	14	13	29	24	13	13	13	12.14	13	11	12								
149	14	13	29	25	10	15	14	14.20	14	11	12	2		1					
150	14	13	29	25	11	13	13	11.14	14	12	12			1					
151	14	13	29	25	11	13	13	11.14	15	12	12								2
152	14	13	29	25	11	13	13	11.14	15	12	12	1							1
153	14	13	29	26	11	13	13	12.15	15	12	11								1
154	14	13	30	22	11	11	13	12.15	14	9	11								1
155	14	13	30	23	10	11	12	13.14	14	10	13			1					
156	14	13	30	23	10	11	12	13.17	14	10	11			1					
157	14	13	30	23	10	11	12	14.19	14	10	13								
158	14	13	30	23	10	15	13	14.16	12	11	14								9
159	14	13	30	23	10	15	13	14.16	12	12	14								5
160	14	13	30	23	10	15	13	14.16	12	13	14								1
161	14	13	30	24	9	13	15	14.15	14	12	11	1							1
162	14	13	30	24	10	11	14	14.18	14	11	12								1
163	14	13	30	24	10	13	14	15.15	14	11	12			1					
164	14	13	30	24	10	15	14	10.17	14	11	12								1
165	14	13	30	24	11	11	13	19.20	14	10	12								1
166	14	13	30	24	11	13	13	16.22	15	12	11								
167	14	13	30	24	11	14	13	15.16	14	11	11	1		1					
168	14	13	30	24	12	13	13	11.14	15	12	13								
169	14	13	31	24	10	14	13	15.18	14	12	11	1							
170	14	13	31	24	11	14	13	13.20	14	11	11	1							
171	14	13	31	25	9	14	13	15.17	14	11	12	1							

Table 1 (Continued)

Ht	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385a	DYS 437	DYS 438	DYS 439	Ecuador [Kichwa]	Ecuador [Mestizo]	Ecuador [Afro-Ecuadorian]	Ecuador [Waorani]
172	14	13	31	25	10	14	13	15.17	14	11	12	2		1	
173	14	14	29	24	9	11	12	12.18	14	10	11		1		
174	14	14	29	24	11	13	13	11.14	15	12	12		1		
175	14	14	30	23	10	11	12	13.16	15	9	12				
176	14	14	30	23	10	14	13	14.15	14	11	11	1	1		
177	14	14	30	23	11	13	13	11.14	15	12	12		1		
178	14	14	30	24	10	13	13	11.14	14	12	11		1		
179	14	14	30	24	10	13	13	11.14	15	12	11		1		
180	14	14	30	24	11	13	13	11.13	14	12	11		1		
181	14	14	30	24	11	13	13	11.14	14	9	12		1		
182	14	14	30	24	11	13	13	11.14	14	12	12		1		
183	14	14	30	24	11	13	13	11.14	15	12	11	1	1		
184	14	14	30	24	11	13	13	11.15	15	12	12		1		
185	14	14	30	24	11	13	14	11.14	15	12	13			1	
186	14	14	30	25	10	13	14	14.16	14	12	13		1		
187	14	14	30	25	11	13	13	11.14	14	12	11		1		
188	14	14	31	23	10	13	13	11.14	14	12	12			1	
189	14	14	31	24	10	14	13	13.18	14	11	13				
190	14	14	32	23	10	14	13	14.16	14	11	12	1			
191	14	14	32	24	10	14	13	13.19	14	9	12	1	1		
192	14	14	32	24	11	13	13	11.15	14	12	11		1		
193	14	14	32	24	11	13	13	12.14	14	12	12		1		
194	15	12	27	24	11	11	12	13.18	16	9	13		1		
195	15	12	27	25	10	13	12	12.19	14	10	13		1		
196	15	12	28	22	10	11	13	13.14	15	10	11		1		
197	15	12	28	23	10	11	12	13.16	15	9	13			2	
198	15	12	28	23	10	12	15	15.16	14	10	11		1		
199	15	12	28	24	10	11	12	14.18	16	9	13		1		
200	15	12	29	22	10	10	14	16.16	13	12	11				
201	15	12	29	22	10	11	13	12.17	16	10	11	1		1	
202	15	12	29	22	11	11	13	14.14	17	10	12		1		
203	15	12	29	23	11	13	13	15.16	13	9	12				
204	15	12	29	24	11	13	13	11.14	15	12	12	1	1		
205	15	12	31	23	12	15	13	12.15	14	12	12	1			
206	15	12	31	24	10	18	13	14.14	16	11	11	2	1		
207	15	13	28	24	12	13	13	11.14	14	12	12		1		
208	15	13	29	21	10	11	14	14.16	14	12	11		1		
209	15	13	29	22	10	11	13	13.14	16	10	11		1		
210	15	13	29	23	9	11	12	12.17	14	9	11		1		
211	15	13	29	23	10	13	14	11.14	15	12	12				
212	15	13	29	23	10	14	13	14.15	14	9	11	2		1	
213	15	13	29	24	11	13	13	11.14	15	12	12				
214	15	13	29	25	11	15	13	15.15	15	11	11	1		1	
215	15	13	30	21	10	11	13	15.16	14	11	11			1	
216	15	13	30	21	10	11	13	16.17	14	11	12			1	
217	15	13	30	21	10	11	15	16.17	13	11	12			1	
218	15	13	30	21	11	11	14	15.17	14	11	11		1		
219	15	13	30	22	9	11	13	13.15	14	10	11			1	
220	15	13	30	22	10	12	13	15.17	14	11	13			1	
221	15	13	30	23	10	11	14	13.15	16	10	11		1		
222	15	13	30	23	12	13	13	14.16	13	9	11			1	
223	15	13	30	24	10	11	12	13.14	16	8	12		1		
224	15	13	30	24	11	13	13	11.14	14	12	12			1	
225	15	13	30	25	11	13	13	10.11	15	12	12			1	
226	15	13	31	21	10	11	13	15.17	14	11	12			1	
227	15	13	31	21	10	11	13	16.17	14	11	11			1	

228	15	13	31	21	10	11	13	16.17	14	11	12						1
229	15	13	31	21	10	11	14	15.19	14	11	12						1
230	15	13	31	21	10	11	14	16.17	13	11	11						1
231	15	13	31	21	10	11	14	16.17	14	11	13						2
232	15	13	31	21	10	11	15	17.18	14	11	12						1
233	15	13	31	21	10	12	13	16.17	14	11	12						1
234	15	13	31	21	11	11	13	16.17	14	11	12						1
235	15	13	32	21	10	11	14	15.20	14	11	12						1
236	15	14	29	25	11	13	14	11.16	14	10	12						1
237	15	14	30	24	11	12	13	9.21	14	11	9	2		1			1
238	15	14	30	25	11	13	13	11.14	15	12	12						1
239	15	14	31	21	10	11	13	15.16	14	11	11						1
240	15	14	31	23	10	11	11	14.16	16	10	12						1
241	15	14	31	24	10	11	14	15.16	14	11	12						1
242	15	14	32	21	10	11	13	16.17	14	11	12						1
243	16	12	28	22	10	11	14	14.15	16	11	11						1
244	16	12	29	22	11	11	13	15.16	17	8	11						1
245	16	12	30	25	11	11	13	11.14	15	11	11			2			1
246	16	13	29	23	10	11	12	12.15	15	9	11						1
247	16	13	29	23	10	13	13	11.15	15	12	11						1
248	16	13	29	23	11	11	12	12.16	15	9	11						1
249	16	13	30	21	9	11	13	12.12	14	10	12	1					1
250	16	13	30	21	10	11	14	15.18	14	11	12			1			1
251	16	13	30	21	10	11	14	17.19	14	11	12						1
252	16	13	30	21	10	12	15	17.20	14	11	12						1
253	16	13	30	21	11	11	15	17.18	14	12	12						1
254	16	13	30	24	9	11	13	12.12	14	10	13	1		1			1
255	16	13	30	24	9	11	13	12.14	14	10	12						1
256	16	13	30	24	11	14	13	11.15	14	12	12						1
257	16	13	30	25	10	13	14	19.19	15	10	11	2					1
258	16	13	31	21	10	11	15	17.19	14	11	11						1
259	16	13	32	21	10	11	13	15.19	14	11	11						1
260	16	14	31	21	10	11	14	16.18	14	11	12						1
261	16	14	31	21	10	11	14	17.19	14	11	12						1
262	16	14	31	22	10	11	14	14.17	14	11	10						1
263	17	13	30	20	10	11	14	15.18	14	11	12						1
264	17	13	30	21	10	11	13	17.18	13	11	11						1
265	17	13	30	21	10	11	14	15.17	14	11	12						1
266	17	13	30	21	10	11	14	17.19	14	11	12						1
267	17	13	30	21	10	11	15	15.18	14	11	12						2
268	17	13	30	21	10	11	15	16.18	14	12	13						1
269	17	13	30	22	10	11	13	16.17	14	11	12						1
270	17	13	30	22	10	11	13	16.17	14	11	13						1
271	17	13	31	21	10	11	15	17.18	14	12	11						2
272	17	14	30	21	11	11	14	17.18	14	11	11						1
273	17	14	31	21	10	11	14	14.17	14	11	12						1
274	17	14	31	21	10	11	14	17.19	14	11	12						1
275	17	14	31	21	10	11	15	17.18	14	12	12						1
276	17	14	32	21	11	11	15	17.18	14	11	12						1
277	18	13	28	24	10	15	13	14.14	14	11	12	1					1

The Waorani haplotypes are printed in bold.

**Table 2**  
AMOVA pair-wise distance based on  $\Phi_{st}$  values between four Ecuadorian and five other population samples;  $p$ -values are shown above,  $\Phi_{st}$  values below the diagonal

	Spain	Colombia, Antioquia	Colombia, Bogotá	Colombia, Choco	Ecuador, Afro-Ecuadorian	Ecuador, Mestizo	Ecuador, Kichwa	Peru	Ecuador, Waorani
Spain	-								
Colombia, Antioquia	0.0172	-							
Colombia, Bogotá	0.0133	0.0018	-						
Colombia, Choco	0.3463	0.2585	0.2742	-					
Ecuador, Afro-Ecuadorian	0.3621	0.2604	0.2795	0.0056	-				
Ecuador, Mestizo	0.0695	0.0200	0.0166	0.2386	0.2265	-			
Ecuador, Kichwa	0.3267	0.2214	0.2169	0.2669	0.2255	0.1378	-		
Peru	0.1888	0.1129	0.1048	0.1923	0.1616	0.0483	0.0247	-	
Ecuador, Waorani	0.4165	0.2900	0.3187	0.3267	0.3161	0.2714	0.0821	0.0840	-

### 3. PCR and typing

Amplification was performed in a Techne thermalcycler, model Genius® and PerkinElmer 9600 thermalcycler, following the manufacturer's recommendations. The 12 Y-chromosome STRs in the Power Plex-Y™ kit were typed with an ABI Prism 310 automated sequencer. Fragment size and allele designation of different loci was determined by comparison with allelic ladders distributed with the kit (Promega Corporation, Madison, USA). The updated recommendations of the DNA Commission of the International Society of Forensic Genetics for analysis of Y-STR systems were followed [7].

### 4. Quality control

The regular participation of the analysing lab (Quito) in the proficiency testing of the GEP-ISFG Working Group (<http://www.gep-isfg.org>) is certified. The population samples were sent to the YHRD for quality checks prior to publication and received the following accession numbers: Ecuador [Mestizo] YA003332, Ecuador [African American] YA003333, Ecuador [Waorani] YA003334 and Ecuador [Kichwa] YA003335. The populations described herein can be searched at [www.yhrd.org](http://www.yhrd.org) [11] by population name, contributor or accession number.

### 5. Analysis of data

Haplotype diversity values were calculated as  $D = N/N - 1(1 - \sum p_i^2)$ , where  $N$  is the population size and  $p_i$  is the frequency of the  $i$ th haplotype [8]. Pairwise values of  $\Phi_{st}$ , an analogue of Wrights  $F_{st}$  that takes the evolutionary distance between individual haplotypes into account [9,10], were calculated to measure genetic distances between minimal haplotypes of 9 populations ( $n = 2096$ ) with the statistical significance determined by a permutation test (10,000 replicates) (Table 2). We used an implementation of AMOVA provided at the YHRD website [11,16]. The DYS389I allele length was obtained by subtracting the shorter allele from the longer allele at DYS389I/II. To illustrate the relationship between populations based on pairwise  $\Phi_{st}$  an MDS plot was created by using the "Population analysis" tools of the YHRD [11,16] (Fig. 2).

### 6. Results and discussion

A total of 341 samples from four Ecuadorian populations were investigated in this study. The total number of different haplotypes is 277 (Table 1). Ninety-nine different haplotypes were observed in the Mestizo sample, 91 in the Kichwas, 97 in the Afro-Ecuadorians and 6 in the Waoranis. Thus, we observed high haplotype diversities in all three major populations ranging from 0.997 to 0.999 but a decreased diversity in the Waorani population ( $Hd = 0.80$ ). Seven different haplotypes were shared between Kichwas and Mestizos, one between Mestizos and Afro-Ecuadorians, and one between Kichwas, Afro-Ecuadorians, and Mestizos. No haplotypes were shared between Waorani and the other populations. To ask whether common Ecuadorian haplotypes are generally common or widespread, we searched the most frequent minimal haplotypes within the actual YHRD 3.0, release 24 [11]. The most frequent Mestizo minimal haplotype in YHRD locus order 14, 13, 29, 24, 11, 13, 13, 11–14 (7/102) is also the most frequent haplotype in the Western European metapopulation (560/16,257). A cluster of 12 closely related minimal haplotypes occurring two times each dominate the Afro-Ecuadorian sample. These haplotypes are shared with African populations or populations with African admixture, but are not generally frequent outside Ecuador.



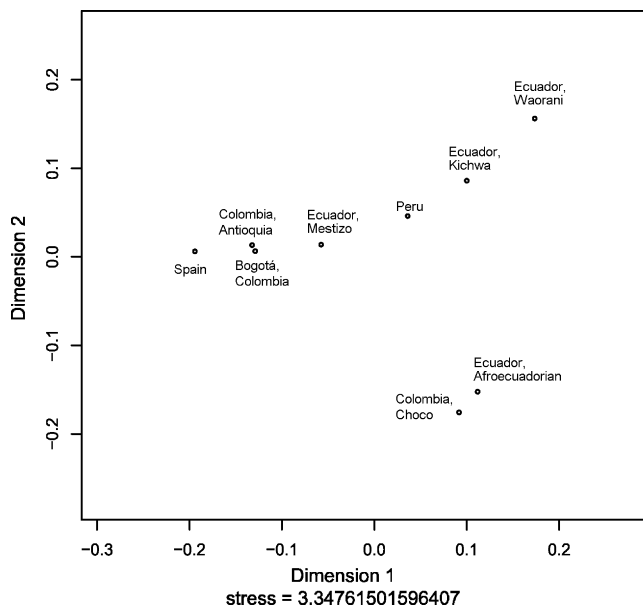


Fig. 2. MDS plot for four Ecuadorian and five reference populations.

The most frequent Kichwa minimal haplotype 13, 13, 30, 24, 10, 13, 14–14 (3/102) has four additional matches only in South America: in the Andean region of Salta in Argentina, the coastal region of Colombia (Nariño province) and in the Wayuu Native Amerindian community of Venezuela. Thus, the common Mestizo haplotypes in Ecuador (including the 1-step neighbours) clearly point to a strong European patrilineal component of this population, whereas the Amerindian and African haplotypes of Ecuador are rather specific for the investigated region. Among the 35 Waorani haplotypes we find only 6 different haplotypes which do not occur in any other of the 499 world populations searchable in YHRD release 24. This shows the strong effect of genetic drift in this endogamous and isolated hunter and gatherer community.

To get a more comprehensive picture of the countries genetic substructure we compared minimal nine-locus haplotypes of the four Ecuadorian population samples via analysis of molecular variance (AMOVA) with several published neighbouring and supposed source populations. These are 777 Colombians of admixed descent from Antioquia province (YHRD accession number YA003087) [12], 147 Colombians of admixed descent from Bogotá (YA003132) [13], 134 Colombians of African descent from Choco province (YA003133) [13], 311 Peruvians of admixed descent (YA003399) [14] and a combined sample of 386 chromosomes from Spain (YA003032, YA003144, YA003303) published earlier as a part of European meta-analysis [15]. The  $\Phi_{st}$  values show significant differences between European/Mestizo, Native Amerindian (Kichwa, Waorani) and Afro-American populations (Table 2). The varying extent to which indigenous and European populations have contributed Y chromosomes to the current gene pool of Western Latin American Mestizos is reflected

by the position of the latter between their source populations in the MDS plot (Fig. 2). As expected, the largest genetic distance exists between the Mestizo, the African and the Waorani samples pointing to a rather strong geographical and socio-cultural isolation. All populations described in this paper have been uploaded to the YHRD 3.0, release 24 [11].

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