Letters to the Editor

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North and South Amerindians May Have the Same Major Founder Y Chromosome Haplotype

To the Editor:

Recently we reported the discovery of a major Y chromosome founder haplotype in Amerindians from South and Central America (Pena et al. 1995). We haplotyped Y chromosomes with two different PCR-based DNA polymorphisms. The first was the tetranucleotide microsatellite *DYS19* (*Y-27H39*), which has at least nine alleles worldwide (Santos et al. 1996) and a gene diversity of .66 in Caucasians (Santos et al. 1993). The other polymorphism, called αh, is a PCR-based system that amplifies divergent units from the alphoid centromeric region (DYZ3) of the human Y chromosome, causing the formation of heteroduplex molecules between products with slightly different sequences (Santos et al. 1995*b*). Together, αh and *DYS19* allow the identification of >45 Y chromosome haplotypes worldwide (Pena et al. 1995; and F. R. Santos, N. O. Bianchi, and S. D. J. Pena, unpublished data). In our initial study we haplotyped 73 Amerindian individuals from 12 different tribes ranging from Argentina to Mexico (Mapuche, Wichi, Chorote, Chulupi, Toba, Huilliche, Atacameño, Surui, Karitiana, Quechua, Auca, and Maia) and identified the presence of haplotype IIA in 74% of them (Pena et al. 1995). We then studied 37 further Amerindians belonging to five tribes from the Amazon Basin and Central Brazil (Waiwai, Gavião, Zoró, Suruí, and Xavante). Again, haplotype IIA was found in the great majority (87%) of the individuals (Santos et al. 1995*a*). By pooling results from the two studies, we calculate that haplotype IIA was present in 78% of the South Amerindians tested ($N = 110$). If we exclude Mapuches, who have a high degree of admixture, this percentage increases to 90%. On these bases we could conclude that haplotype IIA is the major, and perhaps the single, founder haplotype in South Amerindians. We then decided to study North Amerindian populations to ascertain whether the same founding haplotype could be found.

Blood samples were obtained from Mvskoke (Creek) Amerindians in Oklahoma, and DNA was prepared by standard methods. The specimens were collected from volunteer adults in the course of a study of risk factors for adult-onset diabetes. While the Mvskoke include numerous genealogies, the sampled individuals were no more closely related than second cousins. Forty-seven males were typed for the αh and *DYS19* polymorphisms by using our previously described methods (Santos et al. 1993, 1995*b*). The results are shown in table 1 together with a previously published sample of 100 Brazilian Caucasians. The most common haplotype in Mvskokes was IIA (38% of the individuals tested), the same haplotype identified in the great majority of South Amerindians. The second most common haplotype was IIB, followed by IIC, IB, and IIIB. Haplotypes IIB, IIC, and IIIB are very common in Brazilian Caucasians (table 1), while haplotype IB has been seen in other Caucasians (see, e.g., F. R. Santos, N. O. Bianchi, and S. D. J. Pena, unpublished data). One instance of the typical African haplotype IXD (Santos et al. 1995*b*) was also identified among the Mvskokes.

These results become meaningful when they are analyzed in the light of what is known about the history of the Mvskokes (Swanton 1922). They had their first contact with white man in 1541, during the Hernando de Soto expedition. Beginning in the 17th century, European indentured servants began to escape and join the Mvskokes, where their children became full citizens, since the Mvskokes calculate citizenship entirely through the matrilineal line. Soon after, Scots-Irish traders began to reside in Mvskoke towns, marrying the

Table 1

Y Chromosome Haplotypes

Haplotype	Population	
	Mvskokes (n = 47)	Brazilian Caucasians[a] (n = 100)
IA	2 (.04)	2 (.02)
IB	4 (.08)	
IC	1 (.02)	2 (.02)
ID		1 (.01)
IE		1 (.01)
IIA	18 (.38)	10 (.10)
IIB	12 (.25)	38 (.38)
IIC	4 (.08)	8 (.08)
IID		1 (.01)
IIIB	3 (.06)	6 (.06)
IIIC	1 (.02)	5 (.05)
IIID		2 (.02)
IVC		7 (.07)
IVC, Z[b]	1 (.02)	
IVD		2 (.02)
VA		2 (.02)
VB		2 (.02)
VC		2 (.02)
IXD	1 (.02)	1 (.01)
XIIA		1 (.01)
XIVB		1 (.01)
XVA		2 (.02)
XVIA		2 (.02)
XVIB		1 (.01)
XVIIB		1 (.01)

NOTE.—The whole numbers represent the no. of subjects that come from the given population, and the frequencies (in parentheses) represent the presence of that haplotype within the population.

[a] This random sample of 100 Brazilian Caucasians is included for comparison and has already been published elsewhere (Pena et al. 1995).

[b] Two *DYS19* alleles were seen in this individual, suggesting a locus duplication. We have already reported one instance of locus triplication in *DYS19* (Santos et al. 1996).

daughters of prominent Mvskoke chiefs to facilitate the trade in deer skins. By the latter part of the 18th century, the number of whites joining the Mvskoke Confederacy was sufficient for the establishment of two new towns to accommodate them, Big Spring and Broken Bow. African descendants among the Mvskokes have two sources, the so-called "Maroons" (Price 1979) and escaped slaves (Littlefield 1979). The >450 years of intimate contact between the Mvskokes and Europeans allow us to explain the relatively high frequency of Caucasian haplotypes among them as being due to admixture. Therefore, the finding that IIA as the most frequent haplotype at a frequency of .38 is strong evidence that for this North Amerindian tribe this is also a major founder haplotype. Although further studies will be necessary for confirmation, our data support the notion that North and South Amerindians originated from the same single migration of an ancestral Asian population in the Pleistocene.

FABRÍCIO R. SANTOS,[1] LUÍS RODRIGUEZ-DELFIN,[1] SÉRGIO D. J. PENA,[1] JOHN MOORE,[2] AND KENNETH M. WEISS[3]

[1]*Departamento de Bioquímica, Universidade Federal de Minas Gerais, Belo Horizonte, Brazil;* [2]*Department of Anthropology, University of Florida, Gainesville; and* [3]*Department of Anthropology, Pennsylvania State University, University Park*

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Address for correspondence and reprints: Dr. Sérgio D. J. Pena, Departamento de Bioquímica, Universidade Federal de Minas Gerais, Caixa Postal 486, Belo Horizonte, Brazil 30161-970. E-mail: spena@dcc.ufmg.br

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The Gene for Replication Factor C Subunit 2 (RFC2) Is within the 7q11.23 Williams Syndrome Deletion

To the Editor:

Williams syndrome (WS) is a developmental disorder with multiple system manifestations, including supraval-