

ORIGINAL INVESTIGATION

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**T** **a** **3a** **ac** **Ca3ca** **3:**  
**c** **Y-c**

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**Abstract** A recent analysis of mtDNA variation in the Caucasus region in the Iberian, Iranian, and Armenian populations of the Caucasus region. The analysis was based on 11 polymorphic sites in the D-loop region of the mtDNA control region. The results show that the Iberian, Iranian, and Armenian populations are genetically distinct and form a unique cluster within the Caucasus region. The analysis also revealed that the Iberian population is genetically closer to the Iranian population than to the Armenian population. The results suggest that the Iberian and Iranian populations may have a common ancestor who lived in the Caucasus region during the Neolithic period. The analysis also revealed that the Armenian population is genetically closer to the Iberian population than to the Iranian population. The results suggest that the Armenian population may have a common ancestor who lived in the Iberian region during the Neolithic period.

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**I** **3c**

The Caucasus, the region between the Caspian and Black Seas, is a major center of genetic diversity in the world.

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The analysis of mtDNA variation in the Caucasus region revealed that the Iberian, Iranian, and Armenian populations are genetically distinct and form a unique cluster within the Caucasus region. The analysis also revealed that the Iberian population is genetically closer to the Iranian population than to the Armenian population. The results suggest that the Iberian and Iranian populations may have a common ancestor who lived in the Caucasus region during the Neolithic period.

YAP mnt va llt mnan, v r rt r an anay s s f Y-Mr m s ma b-a l l y mar rs n tñ sam s t f Ca Vasan y u l a t n s r y u s y anay z f r mtDNA HV1 s q n y v a r a t n (Nas z an St n n 2001).

## Ma a a

### S b Ms

A t t a f 389 sam s (289 y u i b i an 100 y u i b i swabs) fr nu nr lat ma n x u a s v r y u i b i n tñ f i - l v n s b a t a t y tñ nu s y u l a t n s (Fig. 1): G r a n s (Su tñ Ca Vasan s a rs), Arm n t a n s (In - E r a n s a rs), Az r b a l a n s (U r y s a rs), an Abaz n t a n s, Kabar n t a n s, In - s t a n s, C h i n t a n s, an Dar n t a n s (N r tñ Ca Vasan s a - rs). DNA fr m tñ b i sam s s h a b n u s r y u s y f r anay s s f mtDNA HV1 s q n y v a r a t n an A l n s r t n y m r u s m s (Nas z an St n n 2001; Nas z t a . 2001); m y u i b i swabs fr m a l u n a m a s f r m Arm n a an Az r b a l a n y r y u i b i m r r t n m a s tñ sam s s z f r Y-Mr m s m s t s . Inf r m y n s n t an l n f r m a t n a b u t tñ b r tñ a y f tñ n r an tñ n r s a r n t s an a r a n a r n t s y r b t a n f r m a l n r s . G n m y DNA fr m b i sam s s v a s t r a m b u s n a n I s Q y DNA t r a m n t (O r a R s a r y , B tñ n , Wasñ ., USA) r a y n y n u n a tñ n i - m r f r m tñ (Mant a n t s t a . 1982). DNA fr m y u i b i swabs v a s t r a m b u s n a y n y n u n a s a t u n u t r y u r (M u r t a . 1988). B b i s h Y-Mr m s m s n u y t y m r u s m s (Y-SNP) a t a f r m tñ Su tñ Ca V a s s v r u s f r m 25 S v a n s (G r a a), 25 G r a n s f r m Kaz b (G r a a), 12 L z (Az r b a l a n), an 17 Su tñ O s s t a n s (W i s t a . 2001). A t t n a y b i s h a t a n 21 Az r b a l a n s an 47 Arm n t a n s (W i s t a . 2001), an 63 G r a n s (S m n t a . 2000; W i s t a . 2001), l n t f f r l n a n y r s . M f r m u r sam s s ( a t a n t s h n) an v r n t n n n tñ anay s s t a y l y b i t n tñ r a l t s t f a y n tñ s y u l a t n s . B b i s h Y-SNP a t a (S m n t a . 2000; W i s t a . 2001) f r E r a n , N a r East r n , an C n t r a A s t a n y u l a t n s v r a s l n .

### M I Y a r a n a y s s

T n SNP mar rs r y u s y r r t t b y m r u n E r a n tñ N a r East (S m n t a . 2000) y r y n a n sam s s : RPS4Y (M130), M9, M89, M124, M45, M173, M17, M201, M170, an M172 (U n r tñ t a . 2000 an r f r n y s tñ r n); tñ YAP A l n s r t n y m r u s m s (H a m m r a n H r a t 1995) v a s a s . F r a l SNP mar rs y t M130 (RPS4Y), Taqman (A y B i s t m s) a s s a s y r s t a n . P r i m r s an y a b i r b s y r s t a n b u s n a n P r i m E r s s (V r s n l f r M a y l n t s h P v r P C ; P E B i s t m s). P r i m r a n r b s q n y s a r y n n T a b l . 1. R a m n m s y r r a r a s s m b r - y u s y (M u n t a . 1999) M130 v a s y b u s n a tñ b - m r a s h a l n r a d n r s t r a m n f r a m n t i n s tñ y m r u s m r y u r s m b i s v r (K a s r t a . 2000); v r a s tñ YAP A l n s r t n v a s y a s s m b r y u s y (H a m m r a n H r a t 1995). A n sam s s y r y f r a l mar rs ; n s - y a n y v a s f u n b t y n tñ h r a r a t a r r f m a r s - s m b b U n r tñ t a . (2000) an u r r u s t s . Tñ Y-SNP h a - l o r u n m n y a t r u s h r i s a y r l n t tñ r y n t r y m - m n a l n s f tñ , Y C r m s m C n s r u m (2002). Tñ y i - n t y r l a t n s tñ , f tñ Y-Mr m s m s h a i s s , b s -

### Statist a n a y s s

H a y o r u y r s t a n F s t y a s y r y a t v tñ A n q l n 2.000 (S m n t a . 2000), v a s v a s a s u s t a r y u t M a n t l t s t s f r y r r a t n s b t y n m a t r y s . M i t l m n s n a s a t n s (MDS) anay s s (K u r s a 1964) f tñ F s t y a s v a s y a r - r u t v tñ STATISTICA (StatS f t). P r o a m s l n P H Y L I P 3 . 5 y (F i s n s t l n 1993) v r u s t y n s t r a n b a b r - l n n s t r . 0256 T y (A [ s t y a s .

Y-SNP ɦa 1 ɔru 1 s ln tɦi Ca ɦas s

El 1 yn Y-SNP ɦa 1 ɔru 1 s (Fig. 1) v r fu n ln tɦi Ca ɦas s (Tabl 2, Fig. 1). Tɦi m st fr q nt ɦa 1 ɔru 1 s v r F\*, G\* an J2\*; t tɦi r tɦi fr q n ɦi f tɦi s tɦi ɦa 1 ɔru 1 s was 0.53 0.86 ln a 1 ɔru 1 s v t f r tɦi Dar ɦnans. Tɦi Dar ɦnans ɦa a ɦi ɦi fr q n ɦi f ɦa 1 ɔru 1 s I\* (0.58), v tɦi ɦi s v tɦi r v as fu n at a fr - q n ɦi f 0.1 r 1 ss. Svans v r r 1 u s 1 r r t (W 1 s t a. 2001) t ɦay a ɦi ɦi fr q n ɦi (0.92) f ɦa 1 ɔru 1 s F\*, an Kazb ɦi t ɦay a ɦi ɦi fr q n ɦi f ɦa 1 ɔru 1 s J2\* (0.72). N tɦi r ɔru 1 s ɦa an s n ɦa 1 ɔru 1 s at a fr q n ɦi ɔr at r tɦan 0.5, v tɦi tɦi v tɦi n f tɦi smal sam 1 f 12 L 2 ɦi f r v tɦi tɦi fr q n ɦi f ɦa 1 ɔru 1 s F\* was 0.58. Tɦi Dar ɦnans, L 2 ɦi, Svans, an Kazb ɦi v r r s, n ln ɦi ɦa tɦi 1 v st ɦa 1 ɔru 1 s v r s t ɦi s (0.153 0.652), v tɦi r as f r tɦi tɦi r ɔru 1 s, tɦi ɦa 1 ɔru 1 s v r s t ɦi s was 0.779 0.855.

Tv f tɦi tɦi v mm n Ca ɦas s ɦa 1 ɔru 1 s (F\* an J2\*) ar a s v mm n ln N ar East n 1 u 1 at ɦs L ban s , T r s, S r ɦans (S m ɦn t a. 2000), an Iran - ans (W 1 s t a. 2001), v tɦi av ras fr q n ɦi s f 0.165 an 0.28, r s 1 1 v 1 b t r s n t n 1 v r fr q n ɦi s ln ɦi r (av ras fr q n ɦi s 0.021 an 0.074, r s 1 1 v 1 v 1). Tɦi tɦi v mm n Ca ɦas s ɦa 1 ɔru 1 s G\*, 1 s rar ln ɦi r (fr q n ɦi s = 0.061) an ln tɦi N ar East, ɦas b n r r t n ɦi ln tɦi T r 1 s t ɦi an L ban s ɔru 1 s (S m ɦn t a. 2000). ɦa 1 ɔru 1 s R1\*, v tɦi ɦi s v mm n ln W st r n an C ntra ɦi r 1 s bs r v ln tɦi Su tɦi Ca ɦas s at fr q n ɦi s ɦi ɦi r tɦi n 0.1, v tɦi r as ln tɦi N r tɦi Ca ɦas s, t 1 s abs nt, r n an 1 s. ɦa 1 ɔru 1 s R1a1\*, v tɦi ɦi s bs r v ln ɦi ɦi fr q n ɦi s ln tɦi su tɦi v st M t rran an r ɦi n, East n ɦi r , an C ntra As ɦa, 1 s r s nt at a 1 v fr q n ɦi ln tɦi Ca ɦas s, sm ɦar t tɦi N ar East. Tɦi tɦi r Ca ɦas s Y-ɦa 1 ɔru 1 s v r at 1 v fr q n ɦi.

Tɦi Dar ɦnans, Svans, an Kazb ɦi ɔru 1 s a 1 ar t b u 1 r s v m ar v tɦi tɦi tɦi r Ca ɦas s ɔru 1 s. Tɦi Fst va 1 was ɦi ɦi st b ty n Svans an tɦi r Ca ɦas s ɔru 1 s (av ras Fst = 0.332), f 1 1 v 1 ɦi tɦi Kazb ɦi (av ras Fst = 0.286) an Dar ɦnans (av ras Fst = 0.25), v tɦi r as tɦi av ras 1 ar v 1 s Fst va 1 am n tɦi r ma n n 1 Ca - ɦas s ɔru 1 s was n ɦi 0.047. Tɦi s ɦi ɦi Fst va 1 s, 1 u - 1 v tɦi tɦi 1 v r ɦa 1 ɔru 1 s v r s t ɦi an r u 1 1 mb r f ɦa 1 ɔru 1 s (Tabl 2) ln tɦi Dar ɦnans, Svans, an Kazb ɦi, ar m st 1 1 v tɦi r v 1 t f 1 n 1 v r t f r - rat n ln smal 1 s 1 at 1 u 1 at ɦs.

Tɦi v r r 1 at n b ty n tɦi ɦi ɦi an ɦi n 1 v (av ras Fst) 1 stan 1 s s arat n 1 ar s f Ca ɦas s 1 u 1 at ɦs was n t stat st 1 ar s n 1 f 1 1 ant. (Mant 1 t st: Z = 0.113, P = 0.671). R m va 1 f tɦi u 1 r s (Svans, Kazb ɦi, an Dar ɦnans) r s 1 t ln a v r r 1 at n tɦat was ɦi ɦi r b t st 1 n n s n 1 f 1 1 ant (Z = 0.301, P = 0.134). T r m 1 n v tɦi tɦi r tɦi Ca ɦas s Mu nta ɦs ɦay an ln 1 -

n t/ stan/ f r t/ Su t/ an N r/ Ca /as s s a-  
 rat y , b t n t/ r was statist/ly signif/ cant (N r/ Ca -  
 /as s : Z=0.331, P=0.133; Su t/ Ca /as s : Z= 0.346, P=  
 0.65). M r / r, t/ av ra/ F<sub>st</sub> va/ b t/ n Su t/ an  
 N r/ Ca /as s /u/ at/ ns ( / n/ u/ r s) was  
 0.075, sim/ ar t/ that am n/ N r/ Ca /as s /u/ at/ ns  
 (0.096) an/ that am n/ Su t/ Ca /as s /u/ at/ ns  
 (0.040). T/ r f r , t/ Ca /as s Mu ntains a/ ar n t t  
 /av /ia a t/ Mabl/ n/ n/ n t/ / n t/ /sur / r f  
 Ca /as s /u/ at/ ns; Inst a , / n t/ / r f/ rat/ n/ n  
 small /s/ at/ /u/ at/ ns s ms t/ /av m/ nat/ t/ /  
 / n t/ /sur / r f Ca /as s /u/ at/ ns.

C m/ ar/ s n f Ca /as/ an, E r / an,  
 an N ar East rn Y- /a/ /ru /s

T/ /a/ /ru /y r s/ / n t/ Ca /as s (av ra/ va/ :  
 0.797) /s/ a m s/ t/ as / /as t/ at/ n C ntra/ Asta (av ra/ va/ :  
 0.824) an/ t/ N ar East (av ra/ va/ : 0.769)  
 an/ /s/ signif/ cant/ / / r (t- t st, P=0.024) t/ an t/ /a/ -  
 /ru /y r s/ / n E r / (av ra/ va/ : 0.633). An  
 MDS / t an n / b r- / n/ n/ r bas / n F<sub>st</sub> va/ s  
 (F/ 3A, B) s/ /t E r / an /u/ at/ ns /nt W st rn an  
 East rn /ru /s as /as b n /bs r/ / r /u s/ (S m/ n  
 t a. 2000), /nt C ntra/ Asta /u/ at/ ns /a/ n/ n b -  
 t/ n t/ W st rn an East rn E r / an /ru /s. T/ /  
 Ca /as s /u/ at/ ns ar /nt r m/ n/ /nt N ar East rn  
 /u/ at/ ns.

T/ s /att r ns /av / r/ r b n / n/ r m b/ t/ /ar-  
 v/ s F<sub>st</sub> /m/ ar/ s ns; t/ m an /ar v/ s F<sub>st</sub> va/ f r t/ /  
 Ca /as s /s E r / /s 0.254, /nt r as t/ m an F<sub>st</sub> va/ f r t/ /  
 Ca /as s /s t/ N ar East /s 0.079, /nt /s/ s/ /  
 n/ f/ cant/ / / r (t- t st bas / n av ra/ F<sub>st</sub> va/ s a/ -  
 n/ f / r /u/ at/ ns, P <0.001). On av ra/ , Su t/ /

Ca /as/ an /u/ at/ ns ar m r sim/ ar t/ b t/ N ar East-  
 rn an E r / an /u/ at/ ns (av ra/ F<sub>st</sub>= 0.038, an  
 0.222, r s / /y /) t/ an N r/ Ca /as/ an /u/ at/ ns  
 (av ra/ F<sub>st</sub>= 0.097 an 0.303); /nt /v / r, b t/ t/ Su t/ /  
 Ca /as s an t/ N r/ Ca /as s ar m r sim/ ar t/ t/ /  
 N ar East t/ an t/ E r /, /nt /r s, /t Y-SNP /a/ /-  
 /ru /s.

T/ MDS an F<sub>st</sub> anay s s /n/ s m /ru /s fr m  
 W/ s t a. (2001) /n /nt t/ M201 mar r, /nt /s-  
 t/ n/ s/ / /a/ /ru /G\* fr m /a/ /ru /F\* (F/ 2),  
 was p t anay z (Tab/ 2). In t/ ab /y anay s s, t/ s  
 /n /y u as /v r /ass/ / as /a/ /ru /F\*, a t/ /u /  
 s m u n n /v n r /r/ n /u/ b /a/ /ru /G\*. T/  
 t r m/ n /nt t/ r t/ s /nab/ /t /st/ n/ s/ b t/ n  
 /a/ /ru /s F\* an G\* f r s m /ru /s /n/ n/ t/ /  
 r s/ t/ s f t/ MDS an F<sub>st</sub> anay s s, /v /ass/ / a/ /  
 /a/ /ru /G\* /n /y u as as /a/ /ru /F\* an r / at  
 t/ anay s s. T/ r s/ t/ s (n t /s/ /v n) /v r s s n/ a/ /  
 /nt /a/ ; t/ s, t/ /nab/ /t /st/ n/ s/ b t/ n /a/ -  
 /ru /s F\* an G\* /n s m /ru /s s n t/ /n/ n/ /  
 u r / n/ s/ ns.

In r r t / an/ s s/ b/ blas /n u r / n/ -  
 s/ ns /as b/ t/ small s/ z f t/ sam/ s fr m s m  
 f t/ /ru /s, /v r at a/ anay s s aft r / n/ a/ /  
 /ru /s /nt/ sam/ s/ z /s s t/ n 25 (s Tab/ 2). All  
 /v n/ s/ ns r m/ n t/ sam (ata n t /s/ /v n).

G n t/ /r/ at/ ns/ b t/ n Basq /s an Ca /as/ an s

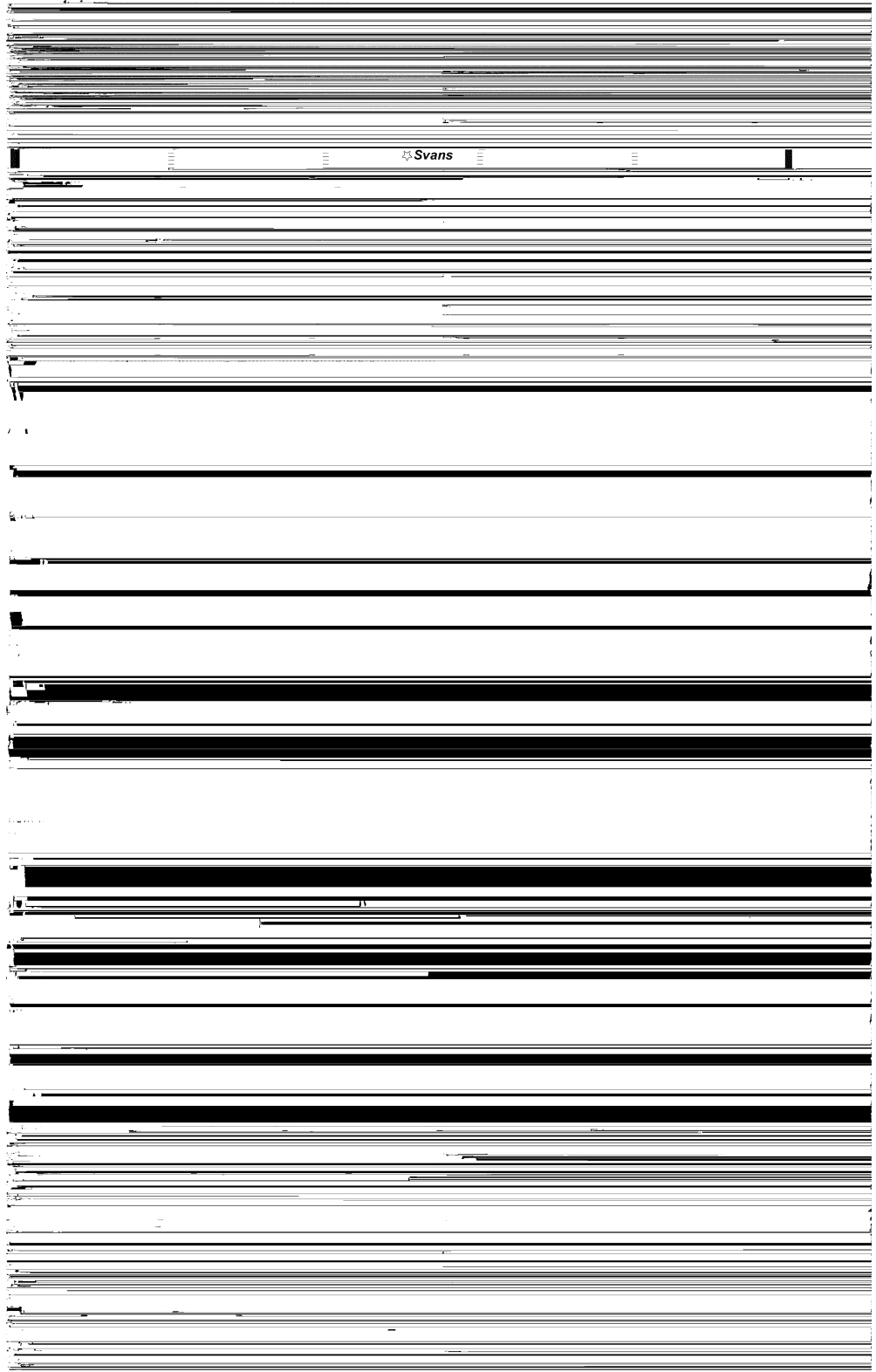
A /mm n r/ n f Ca /as/ an an Basq /s, a r s /as

av ras aryls F<sub>st</sub> va b ty n Basq s an Ca vaslan  
 ru s s m v r (F<sub>st</sub> = 0.563) tñan tñat b ty n  
 Basq s an In -E r an ru s (F<sub>st</sub> = 0.311). Tñ Ca -  
 vas s ru s an Basq s ar n tñ st r t tñ r l n t-  
 tñ r tñ n b r - n n r r tñ MDS t (Fig. 3A, B).  
 Tñ s r s ts ar l n ar m nt v tñ r v l u s t t s bas  
 n "Massa" mar rs an mtDNA HV1 s q n v ata  
 (B r t r v t a. 1995; Nas z an St n l n 2001).

Lan a r v m nts an n t v r l at n s t s  
 n tñ Ca vas s

Tñ r s n v l n tñ Ca vas s, f ru s v tñ s -  
 ra n v n b r s ar n tñ r l n s t n b r s a v s  
 u s t a r s s tñ s t n a s t v n b t t r l a n s tñ  
 n t v r l at n s t s f tñ s ru s: a r a r l an-

**Fig. 3A.** **B** The **Y** SNP  
 relationships of **Y**-**SNP**  
 data from **10**  
**Y**-**SNPs** and the **YAP**  
**A** MDS plot based on  
**F** values and **YAP**  
**S** values among the **Caucasians**, **E-**  
**r** **Iran**, **North East**, and **C**  
**entral** **Asian** **populations** (**open**  
**stars** with **border** **of** **Italy** **popu-**  
**lation** **names** **Caucasians** **are**  
**closed circles** **Iran** **and**  
**YAP** **values** **open squares** **Central**  
**Asian** **are** **closed diamonds**  
**North East** **are** **open triangles**). The  
 stress value for the MDS plot  
 is 0.134. **B** N<sub>0</sub> substitution  
 rates for the same populations  
 (**boldface** **Caucasians** **are** **are**)



The **Y**-**SNP** data from **10**  
**populations** (see **Table 1**)  
**and** **the** **YAP** **values**  
**for** **the** **populations** **are**  
**shown** **in** **Figure 3A**.  
**The** **MDS** **plot** **is**  
**based** **on** **F** **values** **and**  
**YAP** **values** **and** **is**  
**shown** **in** **Figure 3A**.  
**The** **stress** **value** **for**  
**the** **MDS** **plot** **is**  
**0.134**. **The** **Y**-**SNP**  
**relationships** **of** **Y**-**SNP**  
**data** **from** **10**  
**populations** **and** **the**  
**YAP** **values** **for** **the**  
**populations** **are** **shown**  
**in** **Figure 3B**. **The**  
**stress** **value** **for** **the**  
**MDS** **plot** **is** **0.134**.

The **Y**-**SNP** data from **10**  
**populations** (see **Table 1**)  
**and** **the** **YAP** **values**  
**for** **the** **populations** **are**  
**shown** **in** **Figure 3A**.  
**The** **MDS** **plot** **is**  
**based** **on** **F** **values** **and**  
**YAP** **values** **and** **is**  
**shown** **in** **Figure 3A**.  
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**populations** **and** **the**  
**YAP** **values** **for** **the**  
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**stress** **value** **for** **the**  
**MDS** **plot** **is** **0.134**.

Hypertenzivní onemocnění s výrazným kontrastem v tloušťce