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Update of aims population data and test with the genogeographer admixture module

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ABSTRACT

Individuals from Slovenia, Greece, Albania, and Eritrea were typed with the Precision ID Ancestry Panel and included among GenoGeographer's nine reference populations (Sub-Saharan Africa, Horn of Africa, North Africa, Middle East, Europe, South/Central Asia, East Asia, and East and West Greenland). We tested the performance of GenoGeographer with the Admixture Module on AIM profiles of 3548 individuals assumed to belong to one of the reference populations. A total of 3387 (95.5 %) profiles were assigned to one or more of the reference populations, either a single population or an admixture of two or more populations, while 161 (4.5 %) profiles were not assigned to any reference population or admixtures thereof. For 1486 AIM profiles with no reference population of origin in GenoGeographer, the rejection rate was more than 70 % for AIM profiles from North and South America and less than 20 % for those from Central, North, and Northeast Asia.

1. Introduction

The precision of the ancestry prediction using ancestry informative markers (AIMs) is dependent on well-defined reference populations with high-quality genotypes [1]. We have supplemented the previously published European and Somalian data on AIM profiles obtained with the Precision ID Ancestry Panel [2]. Previous studies showed that GenoGeographer rejected more than 20 % of AIM profiles, probably due to genetic admixture of the rejected AIM profiles [3]. We analysed more than 5000 AIM profiles with the GenoGeographer Admixture Module [4] to test if the admixture module decreased the rejection rate without increasing the population assignment error rate.

2. Materials and methods

Individuals from Albania (N = 94), Slovenia (N = 96), Greece (N = 79), and Eritrea (N = 88) were typed for 165 AIMs with the Precision ID Ancestry Panel (Thermo Fisher Scientific). AIM profiles of 5034 individuals from 123 countries were retrieved from publically accessible databases [3]. The data were analysed with the GenoGeographer Admixture Module [4]. The criteria for the population assignment and evaluation of the assignment are shown in Table 1.

2.1. Reference population data

Albania, Greece, and Slovenia were included in the European reference population, while Eritrea was included together with the Somalian reference population to constitute "Horn of Africa".

3. Results and discussion

Of the 5034 test AIM profiles, 3548 profiles were assumed to have a population of origin among the reference populations present in GenoGeographer. The AIM profiles were from individuals from Sub-Saharan Africa, the Horn of Africa, North Africa, the Middle-East, Europe, South/Central Asia, East Asia, and East and West Greenland. A total of 161 (4.5 %) profiles were not assigned to any reference population or admixtures thereof, while 3387 (95.5 %) profiles were assigned to one or more of the reference populations either as single populations or admixtures of two or more populations. Of the AIM profiles with assigned reference populations, 3231 (95.4 %) were assigned to reference populations concordant with the reported populations of origin. In areas with no reference population like the Americas, GenoGeographer rejected the majority of the AIM profiles. However, many of the profiles from Central Asia and Siberia were predicted as genetic admixtures with a Greenlandic component. Fig. 1 shows a world map with the approximate

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Table 1
Criteria for population assignments and evaluation of the assignments.

Observation	Assignment	Evaluation	
		Concordant	Discordant
No population accepted	No population of origin was assigned	Not relevant	Not relevant
One population accepted	The population was assigned	The population of origin was the same as the assigned population (single concordant)	The population of origin was different from the assigned population (single discordant)
Admixture of two or more populations accepted	The admixture was assigned	The population of origin was included in the admixture (admixed concordant)	The population of origin was not included in the admixture (admixed discordant)
≥ 2 reference populations accepted			
One population or admixture with statistically significantly higher likelihood than all other investigated populations	The population or admixture with the highest likelihood was assigned	The population of origin was the same as the assigned population or it was included in the admixture (admixed concordant)	The population of origin was different from the assigned population and not included in the admixture (admixed discordant)
Two or more reference populations and/or admixtures with likelihoods that were not statistically significantly different	The populations and/or admixtures with equal likelihood were all assigned	The population of origin was the same as one of the assigned populations or was included in the admixtures (admixed concordant)	The population of origin was not among the assigned populations and not included in the admixtures (admixed discordant)

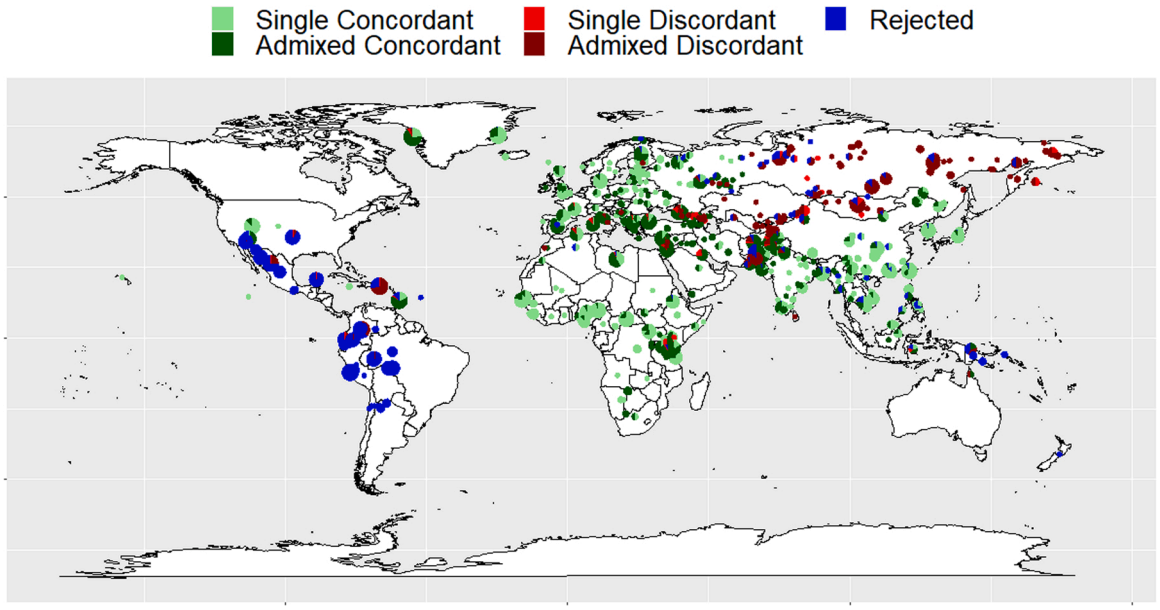

Fig. 1. World map with pie charts showing the results of GenoGeographer analysis of 5034 AIM profiles.

geographic origin of the tested AIM profiles. The pie charts represent the ancestry prediction with the GenoGeographer Admixture Module. In geographic areas represented by the reference populations, the prediction error was low. In geographic areas with high levels of genetic admixture such as the Middle East and North Africa, the GenoGeographer Admixture Module also predicted the genetic admixtures. In areas with no reference population like the Americas, GenoGeographer rejected the majority of the AIM profiles. However, many of the profiles from Central Asia and Siberia were predicted as genetic admixtures with a Greenlandic component.

4. Conclusion

Supplementing GenoGeographer with the Admixture Module decreased the rejection rate without increasing the assignment error rate for AIM profiles when the expected population of origin was included among the GenoGeographer reference populations. For AIM profiles with no reference population of origin in GenoGeographer, the rejection rate was high for AIM profiles from North and South America and lower for those from Central, North, and Northeast Asia.

Conflict of interest

The authors declare no conflict of interest.

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