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Evaluation of the genetic affinity between populations based on the comparison of allele distributions in two highly variable DNA regions

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Abstract. This work compares the allele distributions in the two hypervariable mini-satellite regions (HVR) of the human genome: D7S21 and D12S11 obtained from the Polish population and from other world populations. The graphic analysis based on every 100 base pairs alleles frequency intervals proved the similar distribution of alleles in both the Polish and other Caucasian populations of Europe as well as significant differences in the structure of distributions when we compared the investigated Polish population, representing Caucasians, with Asian population and particularly Afro-Caribbean population.

Keywords: D7S21; D12S11; Interpopulation comparison; Genetic affinity

1. Introduction

Minisatellite DNA consists of tandem repetitive 9–100 base pairs motifs of the length from few hundred to over 20,000 base pairs. These non-coding sequences are the fastest evolving in the genome due to comparatively high frequencies of mutation processes [1]. The investigation of diversity in these hypervariable loci (HVR regions) proves to be a valuable source of information ready to be used to characterise different human race and populations, as well as to define their genetic affinity [2]. The aim of this work is to compare the distribution of alleles in the two highly polymorphic minisatellite DNA regions: D7S21 and D12S11 obtained from the Polish and other world populations. To achieve this the graphic analysis based on the allele frequencies in the intervals of 100 base pair was performed.

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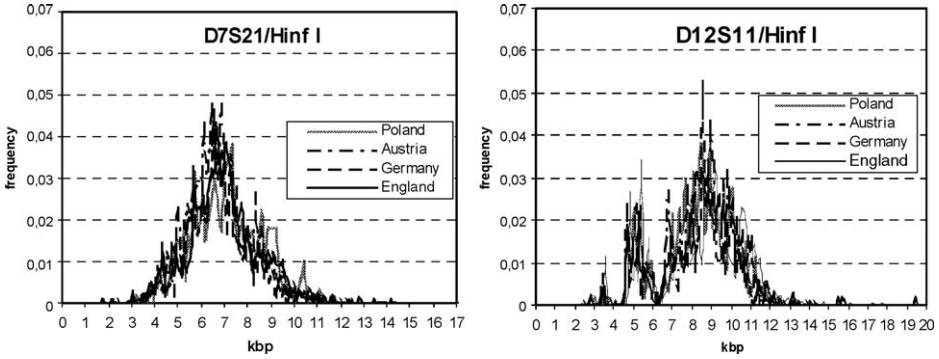

Fig. 1. Comparison of allelic distribution in loci: D7S21 and D12S11 between populations of Europe.

2. Materials and methods

The material used for this research was blood samples of 300 non-related people directly involved in the disputed paternity cases. DNA was restricted with Hinf I enzyme, distributed in 0.7% agarose in TBE buffer, vacuum transfered onto a Hybond N+ membrane, hybridized with alkaline phosphatase conjugating single locus probes: MS31 (locus D7S21), MS43A (locus D12S11) and MW100 (Ladder) [3], detected with the use of Lumi Phos 530 substrate and Hyperfilm™ ECL and analysed with software BIO1D in comparision to the NICE™ DNA Analysis Ladder. The measured DNA fragments were grouped into 100 base pair bins to compare distribution of alleles obtained in the Polish population with analogical distributions for other populations of the world.

3. Results

The restrictive fragments D7S21/Hinf I and D12S11/Hinf I of Polish and other populations of the world were compared on the graphs with the use of size ranges of 100 base pairs. The following graphs contain the comparison of the Polish population with other populations of Europe: Austrian [4], German [5] and English [3] as well as the comparison of the Polish population with the Asian and Afro-Caribbean populations [6] were presented in Fig. 1 and Fig. 2.

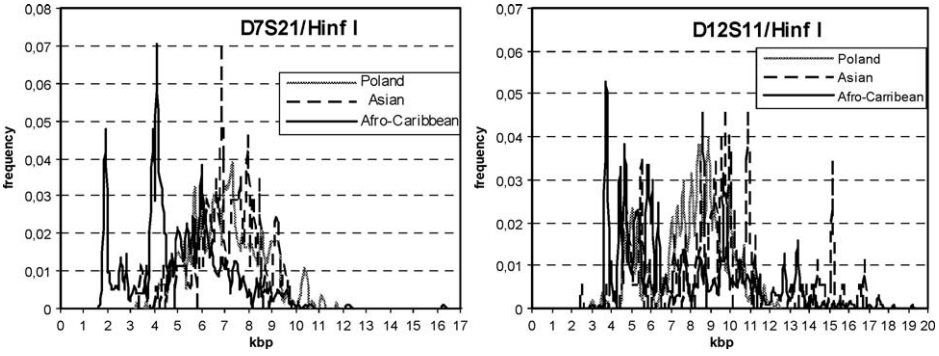

Fig. 2. Comparison of allelic distribution in loci: D7S21 and D12S11 between populations of the world.

4. Discussion

Highly polymorphic regions VNTR are difficult to classify, study and compare due to their lack of discrete, exactly defined alleles. In the HVR locus we observe a high number of variants in the amount of tandem repetitions and a low frequency within population [3]. The graphic analysis based on every 100 base pairs alleles frequency intervals proved the similar distribution of alleles in both the Polish and other Caucasian populations of Europe with characteristic distributions—a mono-modal in locus D7S21 and bi-modal in locus D12S11 [4–6]. Significant differences in the structure of distributions when we compared the investigated Polish population representing Caucasians with Asian population and especially Afro-Caribbean population support the previous observations that the distribution of HVR alleles between populations isolated territorially or represented different human race are quite distinct from each other [7]. The graphs show that the genetic distance separating Caucasian and Oriental populations is statistically shorter than the genetic distance separating Caucasian and Afro-Caribbean populations. As it is presumed by Gill et al. [2] the widest range of DNA fragment occurrence and their highest variability observed amongst the representatives of the Negro race in HVR regions may prove that it is genetically the oldest.

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