Letter to the Editor

Reply to “Bracketing off population does not advance ethical reflection on EVCs: A reply to Kayser and Schneider” by A. M’charek, V. Toom, and B. Prainsack

Dear Editor,

We appreciate the letter by M’charek et al. [1] regarding our publication on DNA-based predictions of human externally visible characteristics (EVCs) in forensics [2]. It indeed was one of the intentions of our paper to stimulate a debate across disciplines by providing scientific facts about the state of the art in this area of biological research, and by developing a scenario for applying DNA-based EVC prediction in the context of future criminal investigations. That our decision to separate the discussion about DNA-based EVC prediction from DNA-based inferences of bio-geographic ancestry (“ethnic origin”) having had triggered the concerns raised by M’charek and colleagues clearly indicates that the underlying scientific knowledge of both aspects is not yet widespread enough. Apart from that, we would like to note that in our previous article we had several reasons not to mix DNA-based bio-geographic ancestry inference with DNA-based EVC prediction intending to deliver the messages on EVCs in a clear and straightforward way. In the following we would like to provide some more explanations, while also including comments on some of the statements made by M’charek et al. in their letter [1].

First, there seems to be a general misconception among the public, including parts of the scientific community, that bio-geographic ancestry in general is externally visible. Although this may be true for some people, it certainly is not true for everybody. In particular, it may be true for those who have most, if not all, of their biological ancestors originating from one and the same geographic region, and that region being one of the few, such as Europe, where people have recognizable homogeneity in appearance traits being distinct from those in people from distant geographic regions. But even for Africa, usually considered as a geographic region that can be assigned based on appearance (and vice versa), this is not completely true since the dark skin color and some facial morphologies usually associated with an African origin are similarly found in Aboriginal people from Near Oceania (e.g. New Guinea), and from Australia. Moreover, being able to infer appearance traits of a person from (e.g. DNA-based) information of bio-geographic ancestry becomes almost impossible in people who are of mixed bio-geographic ancestry having ancestors from different geographic regions. Notably, there currently is no scientific understanding about the relationship between admixture timing and appearance reflection, in other words how many generations it takes until genetic admixture becomes invisible in a person’s appearance traits. Evidence to assume that a person is of “Mediterranean ancestry” (using the example coined by M’charek and colleagues although getting such specific geographic informa-

tion out of DNA is not straightforward) might indeed be helpful during police investigation in search for an unknown person but only if used in a strictly geographic sense. However, it is not clear how “Mediterranean ancestry” information can be helpful when using appearance in the search for an unknown person. In particular, we cannot think of any EVC that is restrictively found in Mediterranean people, but only its existence would serve as prerequisite for such an indirect approach of assuming appearance out of (e.g. DNA-based) bio-geographic ancestry information.

Second, there are principal scientific differences when inferring bio-geographic ancestry, and when predicting EVCs from DNA information. For EVC prediction, particular DNA markers are applied that are either causal of a particular EVC, or are strongly and statistically significantly associated with an EVC, and have proven value for EVC prediction as established from specific statistical analyses of respective genotypic and phenotypic data. DNA markers applied for bio-geographic ancestry testing usually have strong allele frequency differences between geographic regions as established from worldwide population genetic studies. Hence, usually different DNA markers being diagnostic for different kinds of information are used for the two different applications. Scenarios exist where the same DNA marker can be useful for both applications, but this only applies to markers relevant for those EVCs that are geographically restricted, which in fact are rare. One example might be the SNP marker rs1426654 from SLC24A5, a gene assumed to be involved in skin color variation, where the derived A allele is highly frequent in Europeans, whereas the ancestral G allele is highly frequent in Africans and East Asians [3]. However, although there obviously are strong skin color differences between Europeans and Africans/East Asians (having different rs1426654 alleles), skin color also varies between Africans and East Asians (having the same allele), which demonstrates that the relationship between rs1426654 and skin color variation is not yet understood.

Third, M’charek and colleagues are wrong when they write that “EVCs include STRs and SNPs typically located on the Y-chromosome and mtDNA”. Their statement illustrates how the two different issues, bio-geographic ancestry and EVC, can be mixed-up. We do not know of any genetic marker on the Y-chromosome or in the mitochondrial DNA that is related to any particular appearance trait (with the exception of the presence of Y-chromosomal markers in general and male gender determination). Y-chromosomal and mtDNA SNPs are rather informative for inferring paternal and maternal bio-geographic ancestry, respectively. However, performing DNA-based bio-geographic ancestry inference only with suitable Y-chromosomal and mtDNA markers can be misleading, namely in persons of mixed ancestry involving ancestors from different geographic regions. For instance, a “European looking” man might still carry a Y-chromosome of typical African origin in case the paternal African admixture happened many generations ago (and never again since).
Fourth, as much as we see no potential violation of the “right-not-to-know” when it comes to DNA-based EVC prediction, as outlined in our article, simply because appearance traits are known to the relevant person as well as to everyone who has seen this person (or a picture of the person) and therefore cannot be considered private, this is not as straightforward when it comes to DNA-based bio-geographic ancestry inference. Many people may simply not know where most of their biological ancestors came from, and for most geographic regions such information would not be obvious in appearance traits. This is especially true (but by far not restricted) for people of mixed bio-geographic ancestry, see for example the above mentioned “European looking” person with partial African ancestry. Hence, for many people information about bio-geographic ancestry may be new and therefore they may want to keep the right not to know. Special ethical concerns on this issue come from the fact that DNA-based bio-geographic ancestry inference is highly sensitive to reveal non-biological relationship, which in many cases is not evident from appearance and therefore cannot be concluded from DNA-based EVC prediction. Hence, when it comes to ethical concerns, for instance related to the right-not-to-know, DNA-based bio-geographic ancestry testing is more critical than DNA-based EVC prediction.

Fifth, although we entirely agree with M’charek and colleagues that great care needs to be placed in how DNA-based EVC and (bio-geographic) information will finally be used within police investigations, we cannot agree that their scenario of simply concluding Moroccan ancestry from information about Mediterranean ancestry and brown eye color could represent a typical conclusion. Obviously, there is no scientific basis of such a general assumption as there are millions of Mediterranean people with brown eyes who are not of Moroccan bio-geographic ancestry. To understand how significant the problem of population-based preconception is within the investigating body, it would be necessary to establish the relationship of visual perception with ethnic stereotypes and assumptions about potential criminal behaviour in suitable socio-psychological studies among the police forces of European countries, similarly as described for members of the US police forces [4]. If such studies provide confirmed evidence that there indeed would be a significant problem, educational campaigns should be organized to reduce and finally overcome such preconception in parallel with the development of DNA markers for EVC prediction (and for bio-geographic ancestry inference). Furthermore, as it would be in the domain of the police to decide which descriptions for the EVCs/bio-geographic ancestry of an unknown stain donor should be used in their investigation, and whether such information should be disclosed to the public, we would urge to develop a catalogue of unbiased statements for this purpose prior to the implementation of this technology into routine casework to avoid misinterpretation.

Sixth, we cannot see, as claimed by M’charek and colleagues, why there shall be an ethical difference whether EVC information is derived from DNA in comparison to eye-witness reports. In particular, we do not agree with M’charek and colleagues that EVC information obtained from suitable DNA markers is ethically more critical than EVC information obtained from eye witnesses given the arguments they provide. If information from eye-witness reports is considered as “crime-related leads and clues”, which their statement implies, then such an argument can even more reliably be applied to DNA-based EVC prediction because it is performed from crime scene samples, and is based on scientific evidence including information on the accuracy as well as uncertainty of the approach. The fact that only the use of DNA allows to specify a measure of uncertainty of an EVC prediction by means of statistical estimations, which is impossible for information from eye-witness descriptions known to be not very reliable [5], clearly counts in favor of the DNA-based approach over the eye-witness approach. Furthermore, we do not understand why M’charek and colleagues say in their letter that (DNA-based prediction of) “EVC … will in many cases depend on eye-witness accounts to solve crime.” In contrast, we believe that DNA-based EVC prediction can be developed to be sufficiently accurate to either replace often unreliable eye-witness statements, or to be applied in the many cases where eye-witnesses are not available. The example of DNA-based eye color prediction shows that this is indeed feasible [6–8]. Moreover, we find the argument of “genetic policing”, predicting that DNA-based EVC description would turn an entire “ethnic group” into suspects carrying the burden to prove innocence by being forced to provide a “voluntary” genetic sample very difficult to follow, as it would require a deliberate misinterpretation of the data obtained from a given crime stain subjected to DNA-based EVC prediction, and thus far too simplistic. Instead we argue that together with the implementation of scientific progress such as DNA-based EVC prediction and DNA-based bio-geographic ancestry inference (when used appropriately) into forensic practice, care shall be applied via educational efforts to avoid that such new information is used inappropriately. Of course, this needs to be accompanied by an open public debate involving all stakeholders in this process, as well as experts on the ethical, legal, and social aspects. This exchange of letters may be a good start to enter this discussion, and may stimulate others to voice their views as well.

References


Manfred Kayser

Department of Forensic Molecular Biology, Erasmus MC University Medical Center Rotterdam, 3000 CA Rotterdam, The Netherlands

Peter M. Schneider∗

Institute of Legal Medicine, University Hospital, University of Cologne, Melatenguertel 60-62, D-50823 Cologne, Germany

∗Corresponding author

E-mail addresses: m.kayser@erasmusmc.nl (M. Kayser); peter.schneider@uk-koeln.de (P. M. Schneider).