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Wellcome Trust

Nuffield Council on Bioethics Consultation on Forensic use of bioinformation: ethical issues
Wellcome Trust response
30 January 2007

1. The Wellcome Trust welcomes the Nuffield Council's consultation on the important topic of the ethical issues surrounding the use of forensic bioinformation.

2. The Wellcome Trust is the largest charity in the UK and the second largest medical research charity in the world. It funds innovative biomedical research, in the UK and internationally, spending around £500 million each year to support the brightest scientists with the best ideas. The Wellcome Trust supports public debate about biomedical research and its impact on health and wellbeing.

3. Given the Trust's interests, this brief response will focus on the science underlying the forensic use of bioinformation. Consequently, we will only be considering the text and questions primarily related to research issues.

4. However, the Trust is aware of the complex ethical issues that surround the use of DNA databases and funds research in this field via our Biomedical Ethics Funding Programme. Those funded by the Trust in this area include, Mr Robin Williams and Dr Bronwyn Parry, both of whom are members of the Nuffield Council's Working Group.

5. The Trust notes that the composition of the working group does not include a forensic scientist or a representative of the Forensic Science Service. We believe this is relevant because we have some concerns that there are scientific inaccuracies in the text of the consultation document. We hope these will be addressed by engaging with individuals with practical experience of the work of DNA forensics, before the final report is produced.

The science of forensic DNA testing in the UK (p10-11)

6. We would like to take this opportunity to highlight some of our concerns with the body of the text. There is some confusion between changes in technology and changes in nomenclature in the text on page 10 of the consultation document. The phrase "the new genetic technique of DNA profiling superseded DNA fingerprinting" confuses changes in technology with changes in nomenclature. Points (a) to (e) in Box 1 are also confusing in that it appears to relate to the older DNA profiling technology that the consultation document appears to refer to as "DNA fingerprinting".

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7. The first paragraph in Box 1 refers to a "gender marker" which tests for the presence of a small piece of DNA in the amelogenin gene on the male-specific Y chromosome. It tests the biological sex of a sample, rather than the gender, which is a cultural term. It is not entirely reliable: for example, about 2% of men from the Indian subcontinent lack this region of the Y chromosome, but are biologically normal males.

8. The key issues in relation to chance matches and SGM+ profiles are covered by points a) to c) at the bottom of Box 1. However, it is important to note that the SGM+ profiles of identical twins are expected to match and that particular sub-populations may have low variability because of inbreeding.

9. We understand that the debate about SGM+ profiles is being overtaken by developments (particularly in SNP identification and typing) that allow much more detailed information about ancestry and phenotype to be inferred. Please refer to our response in paragraph 13 for a specific example of this.

Question 1: The interpretation of bioinformation
a) In your view, is the SGM Plus sysyem, which uses ten STR markers, sufficiently reliable for use in ascertaining the identity of suspects in criminal investigations and/or criminal trials?

10. It is our understanding that the wording in question one "sufficiently reliable for use in ascertaining the identity of suspects in criminal investigations and/or criminal trials" does not actually reflect the situation in practice. Forensic scientists do not "ascertain the identity"; they report a match and some numbers to quantify the significance of the match, and then it is up to the jury to decide whether or not identity has been ascertained. Given this, we believe the question as framed is not the most valuable question to ask in the circumstances. It might be more useful to ask a question that incorporates the following information: are the numbers used in courts to quantify the significance of a match (a) valid; and (b) comprehensible to jurors? We believe the issue here is not how discriminating is the evidence but whether its discriminatory power is conveyed fairly to jurors.

Ethnic Inferencing (p21-22)

11. It is important to note that DNA can provide information about "biogeographical ancestry" and "demographic background", but not in any direct way about ethnicity because "ethnicity" is largely a social concept. For example, an ethnic classification would distinguish two religious groups living in the same area as separate groups but a classification based on biogeographical ancestry probably would not.

12. We note that the current method of categorisation described in Box 3 ,the seven "ethnic appearance categories", have no accepted genetic validity, and are not genetically distinct from one another. Some, such as distinguishing between "Oriental" and "Asian" are confusing.

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13. It is our understanding that the biogeographical ancestry of an individual can be inferred with high reliability from genetic data if a much larger number of random markers is used, or specific "ancestry-informative" markers are chosen to have large allele frequency differential between population groups. Special panels of markers have been developed for this purpose. From these data, it is possible to infer the pooled continental origin of each of individual's genetic make-up. The reliability of "ethnic inference" about the origin of an individual based on an SGM+ profile is a subject of debate within the research community.

14. In reference to the phrase "the relationship between skin colour and ancestry is complicated and partially determined by social factors", we are unsure as to what specific social factors you are referring to. If you mean that inferences about skin colour may not be robust given "social factors" such as skin and sun bleaching, then we would agree with this statement. It is true that skin colour alone is not a reliable guide to genetic background, and the relationship may be confounded by factors such as sun exposure, however it is our understanding that skin reflection alone is not used to infer genetic background.

15. The phrase: "it is only an inference and does not provide substantive evidence of ethnic origin" is unfortunately worded. Why should an inference be qualified as "only", and why can it not count as "substantive evidence"? Similarly, the comment about "uncertain predictions" skewing police investigations does not seem appropriate. Inferences about biogeographical ancestry will be strong in some scenarios and not in others; the key point is that the strength of the inference can be quantified, e.g. via a likelihood ratio. It is then up to the investigator to take the strength of evidence (the likelihood ratio) into account when deciding where to focus the enquiry. The relevant issue is whether the uncertainty can be quantified in a valid and comprehensible way.

16. In relation to the references you have cited in Box 3, we believe the following additional references will assist the working group in considering this issue. This list includes a Trust funded study by researchers from the University of Leicester that has just been published online in the European Journal of Human Genetics and illustrates the complexity of British genetic ancestry.

- Barbujani G, Belle EM. Genomic boundaries between human populations. Hum Hered. 2006;61(1):15-21

- King TE, Parking EJ, Swinfield G, Cruciani F, Scozzari R, Rosa A, Lim S-K, Xue Y, Tyler-Smith C, Jobling MA. Africans in Yorkshire? The deepest-rooting clade of the Y phylogeny within an English genealogy. European Journal of Human Genetics 2007 (published online on 24 January 2007)

- Serre D, Paabo S. Evidence for gradients of human genetic diversity within and among continents. Genome Res. 2004 Sep;14(9):1679-85

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17. Question 4 b states that: “Certain groups such as some ethnic minorities and young males are disproportionately represented on forensic databases. Is this potential for bias within the database acceptable in law enforcement?” Although a detailed response to this question is outside the Trust’s scope of expertise, we feel it is important to note that “disproportionately” (as in reference to this groups population proportions) does not necessarily reflect any “bias”.