Show me your DNA and I'll tell you your eye color

March 9 2009

More and more information is being gathered about how human genes influence medically relevant traits, such as the propensity to develop a certain disease. The ultimate goal is to predict whether or not a given trait will develop later in life from the genome sequence alone (i.e. from the sequence of the bases that make up the DNA strands that store genetic information in every cell of the body).

Now, writing in the journal Current Biology a group of researchers form the Netherlands put this goal to a test using eye colour. The group around Manfred Kayser of the Erasmus University Medical Center Rotterdam showed that it can be predicted with an accuracy of over 90% whether a person has blue or brown eyes by analysing DNA from only 6 different positions of the genome.

Human eye colour, which is determined by the extent and type of pigmentation on the eye's iris, is what geneticists call a 'complex trait'. This means that several genes control which colour the eyes will ultimately have. Over the past decades a number of such 'eye-colour genes' have been identified, and people with different eye colour, will have a different DNA sequence at certain points in these genes.

Such differences are known as single nucleotide polymorphisms (SNPs). Manfred Kayser and his colleagues analysed the DNA of over 6000 Dutch people whose eye colour had been scored. They determined the sequence at 37 SNPs in 8 eye colour genes for each of these and found that the eye colour of a given individual can be predicted with over 90%

confidence already with the best 6 SNPs from 6 genes, as long as the persons's eyes are blue or brown. For the intermediate colour, shown by about 10% of the people tested, the accuracy is lower at about 75%.

The implications of this study are two-fold. For one, it is a proof-of-principle that complex traits can be predicted from the genome sequence alone, provided that genes with strong effects on the trait exist and are known. This can have implications for predicting disease risks based on DNA, before the disease breaks out. In addition, these findings have direct relevance in the forensic sector. Consider a case where the only trace of the suspect is a DNA trace but the DNA profile generated does not match that of known suspects or any in the Criminal Database.

There currently is in fact one such open case in Germany where the DNA of a single woman was found at dozens of crime sites over several years. Using the approach of the new study, the eye colour of a suspect— and in principle also other traits such as hair colour — can be predicted, thus helping to find unknown suspects. Needless to say, there are also caveats, one of them is that the prediction was only tested for individuals of Dutch European descent, and, although expected, it needs to be shown that similarly high prediction accuracies are obtainable in other populations across Europe.

Also, the reliability of such DNA-based eye color prediction test currently depends on an accurate knowledge that the unknown person whose DNA was tested is of European descent, since the used SNPs are associated with eye color but have no direct functional implications as far as known. Inferring highly accurate information on European ancestry from a DNA sample is not trivial, although such research is underway as well.

Source: Cell Press