Forensic DNA phenotyping: the geneticist’s guide to solving murder mysteries

Here’s a question for you.

Q: If L.A. murderer Blake A. Russell was fleeing the scene of the crime just as the clock was striking 3:10 am on November 23, 2009, bearing in mind that his victim, Sierra Bouzigard, had sustained lethal blunt force trauma wounds to the head; what colour must the perpetrators eyes have been?

A: ‘...You might be thinking, What! That’s not fair! It’s an impossible question! And yet it’s the kind of riddle that police investigators are faced with on a case by case basis. Like any good riddle, it is composed of fragmented statements, or in this case evidence, with no obvious common linking factor. It’d be a pearler if you wanted to stump a brother or sister on a long family road trip, but for criminal investigators such questions are flavoured by the sober task of bringing criminals to justice, and peace of mind to victims and families. So, can the riddle be answered? The answer is yes, and it’s thanks to new and emerging forensic DNA phenotyping technologies such as Parabon Snapshot\(^1\) and Irisplex\(^2\).

Often equated with “painting a picture”, DNA phenotyping refers to the process of predicting what a person looks like from their DNA alone. You see, what L.A. murderer Blake A. Russell didn’t count on when he fled the scene of the crime was that he actually left a little piece of himself behind. A biological witness. By taking genetic material like Russell’s and running it through a predictive model, investigators are now able to produce a mugshot remarkably similar to the photo on your driver’s licence. As you can imagine, this opens whole new avenues within the investigative process. And this technology is not just being applied in new cases-investigators are also digging up cold cases like Russell’s\(^3\) from the deep dark bottoms of filing cabinets pushed nonchalantly into the corners of police stations to see what light DNA phenotyping can shed on these. So how do you spot a killer? Well, it’s all in their expression! Only instead of a dark, harrowing expression behind their eyes, we’re talking about genetic expression.

One Snip, Two Snip, Red Snip... Blue Snip….?

Every trait you possess, from your eye colour to the shape of your nose is controlled by your underlying genetics. The term “expression” refers to how your genetic “recipe” is read and carried out to ultimately result in the product of that recipe. This may be a nose, or perhaps an ear that looks a particular way. Of course, not everyone’s recipes are the same. You can’t make a cake using the recipe for lemonade, and you can’t make detached earlobes using the recipe for attached ones. Crucially, the ingredients that are written in the recipe book determine the taste of the final product. If you substitute eggs where the recipe book says put lemons, the final product won’t be lemonade. In the same way, understanding which instructions in the human genetic sequence, or “recipe book” give rise to which products and how those products “taste” (i.e. look) is a foundational concept for reconstructing a face.

In forensic DNA phenotyping, these variations in the human genetic recipe are referred to as SNP’s.
Pronounced “Snip”, SNP stands for Single Nucleotide Polymorphism. Now, before you say “Whoa, Nelly! You lost me at Nucleotide” let’s break that down. We know that it is true that Single means “only one”. A Nucleotide is simply a “building block” of DNA, with which most of us are familiar from high school biology. Poly is from the Ancient Greek meaning “many”, and Morph means “forms”, which is also Greek. So, what we get is “one building block of DNA that takes on many forms.” The crucial point here is that much like using eggs instead of lemons, having one building block occupying a site on the DNA strand where another could potentially have existed could mean the difference between having blue eyes or brown eyes.5 Snips equal variation, and a unique combination of genetic variations on a face results in what humans experience as “identity”.

AA-Assays and Algorithms

If someone gives you a glass of lemonade, you don’t generally doubt that lemons went into the mix when it was made. After all, you know what a lemon tastes like! Unfortunately, if you are a forensic scientist, it’s a bit more difficult to look at someone with green eyes and be able to tell immediately which SNP in their genome has made them green. To make things just a little more complicated, there are about 10 million SNP’s in the human genome, so finding which SNP links to which trait is a bit like searching for a needle in a haystack. To overcome this, companies like Parabon have taken DNA samples from thousands of people to compile a “library” of genetic information.1 By comparing how many people with a particular trait (e.g. blue eyes) have a corresponding SNP which is similar among them, Parabon have created a formula for the reconstruction of a face based on the DNA of people with “known” traits. Using this formula, the DNA of an unknown person or suspect can then be entered and a prediction (or, “genetic mugshot”) of what they will look like produced.

The crucial word here is prediction, because given the largely complex nature of genetic expression, a genetic profile can never be produced with 100% accuracy of appearance. Let me explain using our recipe analogy again. Lemonade might get its citric taste from the lemons that go into it, but the taste is also determined by the sugar; who’s sweetness serves to balance out the strong acidic taste of the lemon. Then, the whole mixture is weakened by adding water, so that it’s not so strong. Just as it takes the whole combination of ingredients to determine the final taste of the lemonade, a trait on a face is not controlled by just one SNP on one gene. It’s not just controlled by one gene, either. The expression of a trait is determined by a combination of inputs from many SNP’s on many genes; each of which may have varying degrees of control over a trait. Some genes may contribute largely to the expression of a trait, while others may not influence the trait directly at all, or to a very large degree; but may instead impact the gene that does.3 What’s more, some traits, like eye colour, can be predicted using a few select SNP’s contributing largely to expression, whilst others like the shape of a nose are controlled by many more SNP’s, each playing much more subtle roles in expression.7 The result is that genetic mugshots are produced with a degree of confidence; a percentage of likelihood that a person possesses a particular trait.

Also consider into the equation that people alter their appearance, or “identity” through artificial strategies such as hair dye or reconstructive surgery, as it becomes obvious that genetics alone can never create a perfect reconstruction of what a person looks like.

Putting a Name to the Face

For the above-mentioned reasons, cuffing a killer is never quite as easy as identifying the offender from their genetic mugshot in a lineup of suspects. The process is instead deductive, more accurately likened to an elaborate game of “Guess Who”. Traits are ruled out based on what the genetic profile tells us about the offender’s underlying genetics, and the panel flipped on suspects that don’t possess those characteristics.

To exemplify, arresting a suspect on the basis that “It must be Jonny because Jonny has blue eyes like the genetic mugshot” is not really a satisfactory way of arriving at a conclusion because, unfortunately Jean, Harry and Margret also have blue eyes; and they can’t all have committed the crime. Not to mention that there is a small chance that Jonny doesn’t have blue eyes. The margin of error can be narrowed by making statements like, “It’s almost definitely not Bill, because Bill has green eyes and the genetic profile tells us that there is a high chance that the offenders eyes are blue.” In this sense, DNA profiles are designed to produce new leads rather than single out an individual.

In the case of Blake A. Russell, the general consensus had been that Sierra Bouzigard had been killed by a Hispanic male, as she had been keeping in company with Hispanic males on the night she died.7 When the genetic mugshot created using DNA from the crime scene revealed the perpetrator was not in fact of Hispanic descent, the man hunt was diverted to Russell who could then be kept under surveillance. A DNA match was made between the crime scene sample and a sample from an object discarded by Russell, and the case was solved! So, there you have it, forensic DNA phenotyping- how geneticists solve murder mysteries. Oh, and for any one still wondering what colour Blake A. Russell’s eyes were; they were blue. ■

References