DNA Untwisted

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The newest tool for personal identification has gained its biggest and brightest spotlight yet in the shadow of a football legend accused of a brutal double murder. DNA analysis, which claims roots in classical genetics, biochemistry and molecular biology has, from its origins, found itself in an unlikely arena, a court of law. There is no question that the fundamental issues are complicated, but it is possible to present the bottom line conclusion in such a way that a Ph.D. is not necessary to understand its implications. The two most misunderstood buzzwords, which are apparently discussed at dinner tables and cocktail parties 'round the world, are statistics and the C-word, contamination. (I have had people come up to me on mountain tops and ask me to tell them about contamination!). By the end of this piece, you should at least be able to make better cocktail-party conversation.

A Word about Terminology: Fingerprints Come From Fingers

Unreasonable expectations, as well as undeserved criticisms, have been visited upon the entire DNA identification technology of because of the unfortunate terminology, DNA fingerprinting, applied to the original typing method. In its current state, DNA typing is not directly comparable to fingerprints from fingers (dermatoglyphic fingerprints). In dermatoglyphic fingerprints, it is possible to obtain all of the ridge detail information from all 10 fingerpads; thus there are no missing pieces of information. Because only a small portion, perhaps 1 millionth, of the 3 billion units of human DNA are even available for examination by current methods, the result is better compared to a partial fingerprint. Similar to a partial print, however, it may not be necessary to have complete DNA information to be convinced of the individuality of a DNA profile. Just as a certain number of points of comparison have been deemed necessary in order to declare that two fingerprints originated from the same finger, it has been suggested that a defined number of highly polymorphic (variable) DNA loci (chromosomal locations) may be sufficient in order to be convinced that two samples have originated from the same source. One more piece of not-so-trivial information: although identical twins have different fingerprints, in the absence of genetic mutation, the DNA profiles of identical twins are, in fact, identical. More about the DNA of related individuals later.

Another Word about Terminology - Burn the "Match"

Another word that should be banned from the language of DNA typing is the word match. Along with DNA fingerprinting, it misleads the hapless uninitiate into believing that any test called DNA will unequivocally associate a questioned sample with an exemplar. Until all 3 billion of those genetic units can be easily

and reliably analyzed, more appropriate expressions might be the same pattern as,concordant with or indistinguishable from, depending on the strength of the association. The fact that the English language does not provide an easy descriptor of statistical relationships should not detract from the potential power of DNA typing. When many highly variable DNA regions are analyzed, and even the most conservative statistical estimates indicate that not one other person with the same profile exists in the population of the Earth, indistinguishable from becomes one strong statement.

The C word: Contamination

Much of the opposition to the reliability of DNA evidence always seems to return to the now infamous catch-all term contamination. Other than its negative connotation, what does it really mean? Does it only refer to inadvertently introduced material or might it also be applied to a legitimately mixed sample (e.g. blood from two victims). In fact there are a plethora of different types of contamination, and the final, if any, effect on evidence varies. Among the considerations in determining whether a second DNA type would even be detected is the type of testing involved. For instance, PCR-type testing, where the DNA in the sample is copied millions of times over, is inherently a more sensitive technique than RFLP, which also makes a PCR test more likely to detect traces of a second type, whatever the source. In addition, point of view comes into play - one person's contamination is another's mixed sample; it all depends on what you were expecting and for whom you are advocating.

Assuming that the criminalist collecting evidence at the scene isn't bleeding from an open wound, the greatest concern at the crime scene itself is from bacterial, not human, contamination. Crime scene samples, by definition, are in a fertile environment, and fluids like blood and semen provide a very acceptable growth medium for microorganisms. The DNA of the microorganisms themselves is really not a problem - it won't show up in tests that are specific for human DNA. The major concern is degradation of the human DNA in the sample that the bugs are literally using as food. Even so, the DNA type will simply go away, as opposed to being magically converted into someone else's type. Partially degraded DNA must be interpreted carefully by a qualified analyst; if the sample is known to be of poor quality and there is a possibility that part of a pattern has been obscured, a conclusion of "inconclusive" may be the safest bet.

Although great care should be taken as a matter of routine, it is really not that easy to interject extraneous human material into a sample. Contrary to what some might have us believe, DNA does not float around randomly in the air, and cells that may be sloughed off or ejected out of a person are relatively few in number and may not contain any consequential DNA. This is not to suggest that precautions not be taken, but to put the matter in some perspective.

Once the sample is dried, refrigerated and in the laboratory, the potential for contamination is mostly from other samples undergoing processing at the same time. This is where the training, qualifications of the analyst and quality control of the laboratory come into play. Safeguards are set up not only to guard against contamination from other lab samples, but just as importantly, to detect contaminated samples, should they occur. By the way, the criminalist should

remember to wear gloves and not spit in his samples.

The biggest real concern that would actually result in an incorrect DNA type, as opposed to NO type, is a sample switch by the analyst. Until computers can process crime scenes, fully analyze samples and take the witness stand, education, training and good laboratory practice are the best weapons against sample mix-ups.

My Brother Did it

In some DNA typing techniques (not all) a statistical probability is used to estimate the rareness of any particular type - in other words, the possibility that two samples originating from different sources might show the same pattern by chance alone. This type of calculation is valid only with respect to random individuals in a population; it is not applicable to closely related individuals. No two people share the same DNA type except for identical twins. However siblings potentially share more genetic material with each other than anyone else. This is because they inherit their genes from the same two people, Mom and Dad. This idea can be extended to more distant relationships such as children, grandchildren and cousins. In these relationships, some genetic material is shared, but the more distant the relationship, the fewer genes in common. For the highly variable DNA loci that are used in forensic testing, this means that even siblings are unlikely to test the same, especially when many highly variable markers are analyzed. However, until alibis are established all around, your best DNA defense is still "my brother did it..."

DNA in the Judicial System

The statistical interpretation of DNA typing results, specifically in the context of population genetics, has been the least understood (therefore by definition the most hotly debated) issue of recent admissibility hearings. The perceived incomprehensibility of the subject, fueled by the views of, what some feel, have been only a few outspoken individuals, has led to a recalcitrance of the judicial system to accept DNA typing. California, in particular, has become both a hotbed and testing ground for DNA admissibility issues. With some half-dozen conflicting appellate opinions, the California Supreme Court has recently moved to review three recent decisions, and come to a consensus as to whether DNA testing is generally accepted in the relevant community, and may be routinely admitted in criminal trials.