Advances in Forensic Science: Deoxyribonucleic Acid Breakthroughs in the Last Five Years

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Deoxyribonucleic acid (DNA) has come a long way since its discovery by Swiss chemist, Dr. Friedrich Miescher, in the year 1869. It has subsequently been recognized as presenting in three-dimensional, double helix form, has been utilized to exonerate the accused but not-guilty, and has been applied as irrefutable scientific evidence to convict offenders in criminal court, regardless of their lack of admission of guilt. Scientific discovery is a continuum of knowledge to be acquired with each passing year, especially within the field of genetics. The purpose of this paper is to explore what the last five years of DNA research has brought forth, specifically 1) the cost efficiency of focusing on a subset of genomic DNA as opposed to the entire genome, 2) Drs. Phillippy and Miga's establishment of a human genome's length and their estimation of its number of protein-coding genes, 3) Dr. Jiankui's genetic modification of human embryos, and 4) the locating and potential manipulating of genes that are found to be contributory to psychological disorders and criminality.

The American criminal justice system is increasingly turning to DNA to implicate or exonerate potential culprits and to identify remains, so "innovation in genome-sequencing technologies and strategies do not appear to be slowing" (Wetterstrand, 2021). Because there is such a demand, profits are made via volume, as costs continually decrease. In the year 2001, the cost of sequencing a whole genome was a whopping $100,000,000, but by the time the 2014 ended, the cost decreased drastically to a mere $10,000. Following this trend of consistent decline, as of 2022, just last year, the price has been brought down to approximately $1,000 to sequence a whole genome. "The key factors to consider when assessing the 'value' associated with an estimated cost for generating a human genome sequence - in particular, the amount of the genome (whole versus exome)" (Wetterstrand, 2021). Fees could be brought down even

more by focusing on a subset of genomic DNA as opposed to the entire genome. “It is usually sufficient to sequence only a subset of genomic DNA representing regions found to be most variable among individuals. This approach circumvents the much higher cost of sequencing and data management for whole genome sequencing, while providing sufficient specificity for forensic purposes” (Eissenberg, 2018). While this may seem like a short-cut of sorts, sequencing subsets of genomes has proven, in the last five years, to provide all of the information required by forensic scientists to rule-in or rule-out individuals charged with violent crimes without breaking the bank for the State’s sake.

Becoming more cost-efficient while maintaining the quality and integrity of analysis is not the only highlight in DNA’s last five years. Besides this, a computational biologist named Adam Phillippy joined with Karen Miga, a geneticist to become founding members of what they called, The Telomere-To-Telomere Consortium, in an effort to “complete the genome” (Zimmer, 2021). The motivation behind the project was simply to please the perfectionists within them. Put simply, the missing, unknown, undiscovered gaps within the human genome simply annoyed Phillippy and Miga. “They were just really bugging me. You take a beautiful landscape puzzle, pull out a hundred pieces, and look at it — that’s very bothersome” (Zimmer, 2021). The researchers formed a team and worked virtually through the pandemic. The Telomere-To-Telomere Consortium members coordinated and shared data via Slack, a messaging application. When all was said and done, the team of scientists “added or fixed more than 200 million base pairs in the reference genome. They can now say with confidence that the human genome measures 3.05 billion base pairs long” (Zimmer, 2021). During this process, The Telomere-To-Telomere Consortium managed to discover over two-thousand new genes including one-

hundred-fifteen of which are able to produce proteins. Perhaps researchers of the future will be able to identify the function of the proteins produced by the one-hundred-fifteen newly-found genes. As if that was not enough achievement within one project, the team is now also able to "estimate that the human genome contains 19,969 protein-coding genes" (Zimmer, 2021). The fact that all of this was accomplished by researchers involved in telework is a credit to Drs. Phillippy and Miga. What they achieved remotely during a pandemic could not have been achieved by lesser scientists working in-person within the confines of a laboratory.

Another DNA-related breakthrough came toward the end of the year 2018, when a team of Chinese scientists, headed by Dr. He Jiankui, broadcasted that they had utilized CRISPR-Cas9 to genetically modify human embryos. CRISPR-Cas9 "edits genes by precisely cutting DNA and then letting natural DNA repair processes to take over" (CRISPR Therapeutics, 2023). These modified embryos "were then transferred to a woman's uterus and resulted in the birth of twin girls—the first gene-edited babies. The twins' genomes were modified to make the girls more resistant to HIV, although the genetic alterations may have also resulted in unintended changes" (Bennett, 2019). Dr. Jiankui and his team of geneticists were broadly frowned-upon by fellow scientists due to performing a procedure that could be viewed as violating ethical codes, and bringing forth hazards. Many scientists the world over stand by the concept of "just because you can, doesn't mean you should", and because of this, there was a presentation regarding more stringent regulating of how CRISPR-Cas9 and other new tools are utilized, "particularly when it comes to changing the DNA of embryos and using those embryos to birth live children" (Bennett, 2019).

While perhaps Dr. Jiankui opened a door he shouldn't have, due to the unethical and risky actions he took, one cannot deny that his project brought forth major questions regarding the future of humanity. Could the locating and manipulating of genes that are found to be undesirable be a way to better members born into a population, and in turn, improve societies of centuries to come? Geneticists have delved into inheritance in school-aged pairs of twins, noting that "traits of being callous and unemotional combined with antisocial behavior are highly heritable" (Viding, et. al., 2020). Naturally, the youngsters who were deemed as unfeeling and detached bore a higher likelihood in having conduct problems in school settings. Additionally, other researcher teams studied data on partakers in a study that had tracked sibling sets for two decades, from the newborn stage to the young adult stage. This team of research scientists concluded that "showing an early disregard for others in toddlerhood was statistically predictive of specific psychopathy traits in young adults" (Viding, et. al., 2020). In a research study from 2019, Finnish scientists "looked at the protein expression in neurons and astrocytes, comparing psychopaths to non-psychopaths" (Tiihonen, et. al., 2020). Fascinatingly, this was accomplished with the sampling skin cells from violent inmates, non-violent inmates, and non-imprisoned individuals. The team utilized the sampled skin cells to generate stem cells, and then distinguished those stem cells into neurons and astrocytes (brain cell types). Finally, the team of geneticists from Finland identified the genes that were "expressed differently in violent offenders' neurons compared to control subjects. The results showed that ZNF132 was markedly upregulated in the violent offenders, and CDH5 was markedly downregulated. The estimated heritability of severe antisocial behavior disorder is about 50%" (Tiihonen, et. al., 2020). Another study, by Andreas Meyer-Lindenberg, Director of the Central Institute of Mental Health in Mannheim, Germany, revealed that Brunner Syndrome, a rare genetic disorder, is caused by

an MAOA mutation leading to MAOA deficiency, and in turn an excess of monoamine transmitters. Dr. Meyer-Lindenberg established that this causes extreme "impulsive behaviour including hypersexuality, sleep disorder and extreme mood swings as well as a tendency to violence" (Hunter, 2020). This study also revealed that has discovered that "males with this variant had neurobiological structural factors that would predispose them to violence" (Hunter, 2020). Will geneticists of the future follow Dr. Jiankui's lead and activate and/or deactivate genomes of human embryos to rid the planet of psychopaths, sufferers of Brunner Syndrome, and those who would otherwise be affected by genetic-based antisocial and/or violent behaviors? It is indeed a possibility.

The last five years in DNA exploration have brought forth a plethora of breakthroughs, namely, the extremely less costly method of examining subsets of genomic DNA rather than entire genomes without compromising integrity and reliability, the deeming of the length of a human genome and the approximation of how many protein-coding genes are present, the genetic modification of human embryos, and the potential to locate and manipulate genes that are found to be objectionable so that only genetically non-violent humans can be born. Because of these newfound highlights, DNA studies will continue to be a growing specialty among forensic scientists the world over.

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