The Reality of the Equity in Genomics

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The aim of this paper is genomics science and its relation to equitability, which it is not equitable. Genomics research truly has revolutionized our understanding of human biology allowing for tailored treatment, but genomics data and advancements definitely are not proven equal with different groups, always favoring European descent. It exacerbates health inequities as no access and less development in precision medicine for minority groups are a problem. Diversifying genomics research efforts have gone underway but many challenges still stand in the way of equity in genomic healthcare and technologies. Genomics has also been neglected to a proper examination on why its pitch in the world has been neglected on a policy level and in areas that glue society like law, medical and technology fields. It also still is a relatively unknown career space. Using different forms of literature and navigating different genomic websites, we show real problems and solutions regarding genomic science that link to forensic databases, and race-based medicine. Some of the biggest findings involved figuring out that forensic databases' past security has led to a lot of distrust which is dangerous when dealing with a science like genomics. Another big finding was that AI combined with genomic science and genomic tech could help law enforcement with analyzing crime scenes deeply which is in the great interest of a lot of people as it heavily benefits society.

Introduction

Genomics science is the study of genome structure, function, evolution and use. Scientists study the interactions between genes and the environment, as well as how tools from the genome can be used across fields, a prominent recent example being CRISPR technology in medicine for gene editing purposes. CRISPR is absolutely crucial for the development of genomics science, because it can be used for problems like mutations in the human genome that cause issues for humans. One example is a loss of function mutation that leads to cystic fibrosis that can be edited in the genome by using CRISPR technology that results in regaining the function of the ion channel. With its unique DNA cleaving mechanism, CRISPR can help get rid of the mutation and sequence in a proper fixed up gene for that human. Today, scientists can easily modify and regulate genomics loci in a wide array of cells making genomics science especially valuable in the medical field. The knowledge of scouting the human genome also allows for better disease control and surveillance too, so alongside CRISPR, doctors can accurately pinpoint the problem where genomic tools like CRISPR can help with treatment. Tailored treatments can come with individual genetic makeup thanks to these innovations in genomics which can revolutionize potential care but unfortunately, even though genomics innovations like CRISPR have high potential, they also have broader social and political ramifications. The integration into clinical practice raises concern for equity as there are inherent limitations which perpetuate disparities in healthcare. AI's whole decision making process alongside the lack of transparency exacerbate distrust amongst marginalized communities which further serve to widen the chasm when it comes to genomics healthcare.

Beyond CRISPR, there are many contemporary public policy debates around genomic science including governance and regulations around race-based medicine, medical biobanks, germline and somatic cell editing, forensic databases all discussed in Hoschild's Genomic Politics. The pursuit of equitable and proper healthcare with genomics included has the journey with many obstacles rooted in historical legacies of bias, socioeconomic factors and different scientific limitations. The persistence of racial based medicine which in this paper, I will be focusing on alongside AI based precision medicine, perpetuates outdated paradigms tethering medical interventions to socially constructed notions of race, overlooking genetic diversity across populations. Race based approaches serve as proxies for socioeconomic factors, obscuring the cause of inequities. I will be looking into race-based medicine and AI based precision medicine further in depth as they both have very different problems, but similar solutions. Through a multidisciplinary lens encompassing bioethics, social justice, genomics science systems,AI this paper will walk the line of endeavoring the intricacy of the current word of genomics science involving technological advancements,historical legacies and systematic discrepancies. In particular, I will begin with a literature review of contemporary policy issues in these areas and then summarize some proposed policy solutions to hopefully pave the path for a more equitable and inclusive future for genomics science and precision medicine.

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Case Studies

Case - I: Forensic Databases

Forensic databases are databases that store collective genetic information. These forensic databases have multiple uses. One is for storing DNA samples from suspects at a crime scene, which helps law enforcement determine if the DNA of the suspects matches any DNA that might be present at the crime scene itself. With concrete evidence, it would simplify many cases, making forensic databases extremely valuable. DNA can also be taken from previous criminals, which is very useful specifically for unsolved cases. The purpose of pulling up previous if a police officer ever would run into the criminal, they can pull up the database to compare DNA just like the New York city Police department did back in 2000 (Zurr & Catts, 23)[1]. After the genetic information in these databases are derived from DNA samples, DNA sequencing is used to generate DNA profiles in these databases. Forensic databases are especially useful in law.

However, there are issues with forensic databases. Genomic data containing private information about individuals and even their relatives if obtained from a family history or genealogy archive can be leaked. One example of this comes from a family history site called GEDmatch, who on July 22nd 2020 inadvertently leaked 1.4 million individuals' genetic information after it had helped solve a criminal case called the Golden State Killers Case (Murphy 2020)[2]. The DNA Diagnostics Center is another organization that had national genetic testing information that was crucial for development in technology (DNA Diagnostic Center Fined $400,000 for 2021 Data Breach. 2023)[3]. Its information was hacked from May 24th to July 28th 2021 Individuals and their relatives' full names, social security numbers, bank accounts were taken.

Forensic databases have had security breaches before and can result in the theft or misuse of sensitive forensic data like the US Office of Personnel Management had a breach exposing personal information especially the fingerprint records of over 21 million people. Also people with authorized access using the databases for their own gain like an employee who had clearance to the Alabama Department of Forensic Sciences and he tampered with evidence in the state's DNA database (OPM.gov 2023)[4].

Another issue with forensic databases is public skepticism and distrust over its usage in the public domain. Jennifer Hochchild[5] in her book Genomic Politics, talks about how skepticism builds when there is the knowledge of human error at the back of the mind. Forensic databases require care for extracting and storing genetic information and other material, but it is very hard to validate whether it got stored safely. So, doing it for many people at once, will result in not just human error happening, but also errors on inputting information on the databases part.

Genetically driven identification is constant so when forensic databases are not really safeguarded and assumptions start to be made with whole racial groups with testing there starts to be issues. An assumption being like heart disease is attributed to the black population more (Gravlee 2020)[6]. Moreover, conservative and liberal states are debating how to view and use genetic test results and also regulate the usages of forensic databases and biobank activity.

Lastly, American society as a whole is not highly knowledgeable about the wide range of genetic applications for conditions like flu, sickle cell disease, cystic fibrosis, heart disease, as well as genetic and nongenetic causes of varying health patterns. The SNP selection in the genome of the individual varies and its association with the disease can be influenced by genetic and environmental factors (Genetics and Health - Genes, Behavior, and the Social Environment 2023)[7]. There is constant confusion on what genomics can do in the context of assisting our lives in America's rapidly changing society as Hoschild found from her survey in her book Genomic Politics.

She gives the example of gene editing, where there is way more support for it than forensic databases, but people know surface level knowledge on the potential it has to help people on a mass scale. People against gene editing just often see it as a prophecy meets one person or group situation. This is similar for many more topics under genomic science ultimately leading to heavy division in the perspective of Genomics and consensus in American Society.

Case - II: Race Based Medicine

Race based medicine is a term used in the medical industry where race plays a contributing role in the prescription of medicine. The term itself generally means race is used as a biological and genomic marker when giving a health status report to a person. There is a deep history when it comes to how race-based medicine originated to have multiple facets now in our modern medical society. It started with the fact that the human genome mapping technology immediately jumped to asking questions of health disparities between people.

The practice of race-based medicine is an issue since race and ethnicity are terms carrying complex meaning, reflecting culture, history, socioeconomics and more rather than straightforward biological categories. Ancestral origin has some correlation with genetic variation closely related to patient health, so on this fact, race correction started to come. As tools like DNA based group homogenization or DNA markers develop, selection of groups for specific genetic trends start to become more accurate and common in the medical field. Some of these DNA markers that help with ancestry correlation to just straight genetics are Y-DNA markers/Y- SNP markers and x chromosome markers. With the Y-DNA markers specifically it could help with finding Native American lineages with far less recombination possibility from knowledge of past history. This leads to race correction (Mersha & Tilahun,2021)[8].

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Unfortunately, with these developments a problem arises where the correlation of genetics with health is often weak or too specific. One massive issue is that doctors often oversimplify the genetic variations within racial groups, so inaccurate assumptions about people's health are made. Thus, this could potentially lead to devastating under/over treatment. Race does also not have a clear genetic definition really so this can lead to a misinterpretation of genetic data.

To take a recent example, in 2020 doctors believed that African Americans were more vulnerable to the infectious diseases Covid-19 because of an inherent genetic predisposition alongside unmeasured and certain genetic factors. The predisposition was small, but definitely evident. At first the thought was that minorities like blacks were more likely to have Covid-19, because of genetics, but the reality was that since around thirty nine percent of African Americans live in non- suitable living conditions, alongside social factors like racism, minorities like African Americans are more likely to get Covid-19. Unsuitable living conditions include a dirty environment and often extreme poverty. Social factors were more of the reason why the data was showing that African Americans were getting Covid-19 more frequently. Society as a whole was viewing race groups and Covid-19 treatment too literally as different options. Instead the better thing to look at would be the environment for people like food, and air (Gravlee 2020, Tong and Artiga, 2021)[6]. However, the biggest misunderstanding comes from the fact that there is an innate practice of taking race as a medical factor, through teaching which is training the future doctors and medical specialists to think this way. These beliefs keep undergoing the research of a person in the medical field, forming premeditated solutions for specific scenarios of who the patient is and how critical their health problem is.

Another present issue with race-based medicine is that treatments may become too specific for one group of people based solely on racial characteristics, and may fail to address a problem for broader swathes of patients. One example is congestive heart failure in African Americans that the heart medicine BiDil or isosorbide dinitrate, first rolled out in 2005 was designed to solve. The caveat, however, was that this was approved for only African-Americans, and with no control group in the clinical trials. two problems became evident (Smith 2020)[9]. One was that for the multiracial societies all across America, that meant that there was not a medicine that could work for everybody as it was only used for African-Americans, so people besides that racial group would still struggle with congestive heart failure. Second, because there was the single heart medicine BiDil, no other was made that was of BiDils quality, so if you could not take the medicine, there was still no alternative. Medical decisions should not be based on a factor like race.

Policy Reforms

Public Education

One possible public policy reform to address the challenges across the issues raised in this The paper is to increase public awareness of genomic science.

In particular, public education could take the form of a nationwide overhaul of science curriculum.

To help solve the race-based medicine problem, change has to begin with the education in medical schools that reinforce beliefs about race and medicine. This is already underway: in 2020, the American Medical Association is adopting new policies to recognize race as a social construct so this discourages doctors conflation of race as a biological category when interacting with patients. The AMA also funded medical programs to make curriculum changes recognizing how race was dangerous as a proxy for biology in medical education]. Education changes and more widespread access to information would also allow for racial minorities to have more representation in the genetics field, and help steer doctor-patient navigations away from harmful race-based practices.

In 2021, Australia introduced curricula into their public education system so that students, especially 16-18 years old, could develop genomic literacy. Genomic literacy is where people have sufficient knowledge and understanding of genetic principles for individuals to make decisions for participating in social discussions on genetic issues. This curriculum included contemporary application of genomics, conceptual knowledge, and recognition of moral and scientific flaws in genomic research.

School Science curriculums should include the basics of genomics, like genetic variation,gene expression,sequencing technologies,machine learning algorithms and neural networks which will help students realize the benefits and limitations of AI and genomics in different fields. Encouragement of critical thinking when it comes to ethical, legal and social implications of genomics alongside when it comes to data analysis will be important in strengthening the decision making process when it comes to different genomics situations and how to react to it. It also is hands-on experience working with real genomic datasets. Lastly, case studies on how genomics and AI are revolutionizing patient care, alongside the inherent problems that come up with different genomic technologies and its risks are important to note (The impact of AI on School Genetics and DNA studies. 2023)[4]. Medical school curriculum should integrate genomic medicine and AI driven support tools into coursework and clinical training experience prepping doctors to make evidence-based care.

Public education has helped in previous health science contexts as the case of tobacco-smoking in America has shown through campaigns from TIPS, which is a campaign containing tips from previous smokers' campaigns. When ads were made

to help adults reduce smoking, it had a huge impact on the following generation. As kids often look up to adults, when adults reduced smoking, the kids took notice and in 2014 the kids also reduced smoking as well. Anti-smoking media and messages in education portraying negative effects of smoking early on also helped to reduce tobacco usage (Public Education Campaigns: Why They Are Needed - Campaign for Tobacco 2023)[10].

Lastly, education can help American society be more open and knowledgeable about how to help protect their information in these forensic databases for law. Right now, genealogy and forensic databases get its data from crime scenes and direct to consumer DNA testing, which is then used for law and forensic analysis. The direct to consumer DNA testing to then put in the databases alongside forensic DNA data from the scenes helped catch the golden state killer.

DTC genetic tests and genomic technology called SNPs helped relay information that was unique over the six hundred thousand sites in the genome accurately. Then when it was put in the forensic databases, and analysis was run, the forensic databases were able to match the DNA from previous crime scenes with these new genetic tests to figure out the killer with the help of genealogy databases as well. Genome data has features like association with traits and family relationships and with how widely public it is with these databases, its value is high. With this in mind, forensic databases containing genomic data have to be regulated and private in the correct hands is important, as otherwise it can get manipulated to show different results.

Another problem is that investigators with warrants could access these entire forensic databases which contain sensitive genetic information. These are two serious ethical issues and currently less attention has been given for creating policies regarding genomic data. Education would allow for people to first know how valuable genomic data is and what it is used for in law, because awareness helps to clear up any misconceptions. It would also start letting people have the capability to protect and control how their DNA data is used and shared, for these forensic and genealogy databases. Third, it would bring up a discussion on how this technology can remain balanced while letting criminal investigators do their job effectively. For policies, it would let the public have a voice in advocating for proper policy on keeping this genomic data safe because on a large scale, there has to be a policy to ensure protection against threats trying to leak or manipulate this information. NIH has started by helping spread the public's voice and opinions along with bringing the community together to make a proper genomic data sharing policy (NIH Genomic Data Sharing Policy: NIH Request for Public Comments. 2022)[11]. Some real life examples include the NIH subsection being the NHGRI launching the "Genome Unlocking Life's Code Exhibition", where NHGRI collaborates with Smithsonian Institutions to get this exhibition running, and they travel to different museum and scientific centers across the US (Genome exhibition pieces now on display at NIH, 2023)[7].The history, science of genomics and impact on healthcare and society is explored, through displays,multimedia presentations increasing awareness on the potential implications that genomics has for personalized medicine and ancestry relations to genomics. Genetics and Genomic Career resources is something they provide as well for students interested in pursuing careers in these fields fostering interest at an early age. National DNA Day celebrated on April 25th in the USA celebrates the Human Genome Project in 2003 and the educational activities,lectures and engagement allows for more people to be educated on false and true genomic concepts for our society promoting understanding among audiences. Even direct to consumer genetic testing while being often oversimplified raises awareness about genomic ancestry through its campaigns.

AI-Based Precision Medicine

Another possible public policy reform to address the challenges across the issues in this paper is to use precision science or AI to accurately assess a patient's condition without using race as a factor in medical decisions. AI could be used to analyze the genomic data of a person and the doctor could be used to examine health problems with the help of precise AI tools. For example, IBM Watson for Genomics is an AI platform that analyzes genomic data to identify potential cancer-causing mutations and can analyze vast amounts of data alongside clinical trials to make more informed decisions tailored to an individual's genetic profile.With this in mind, the doctors would still have to know certain genes in the genome, depending on where the doctor specializes. One way that the AI would be able to examine and evaluate different health problems is to run the AI on pathological specimens in order to train the algorithm, or neural network. DNA sequencing technology can be used to read out long sequences of DNA. This would allow medical specialists to detect complex variants that are not missense variants, that could link to making a classification for the disease/health issue allowing doctors to act on this interpretation of the genomic data and their own observations of the patient. "PrimateAI-3D,a three dimensional convolutional neural network for variant effect prediction,trained using primate variants and 3D protein structure"(Vuksanj,2024)[12]. International efforts can come through genomics just like with PrimateAI and larger scale projects identified a host of missense variants, creating a catalog of them from 233 primate species. Comparing this to the pathogenicity of rare variants from biobanks is an example of using AI to gain information regarding variants with primitive qualities of humans using a neural network to separate how to fix different patients' problems. (Vuksanaj,2024)[12].

Right now, a problem with the DNA sequencing technology is that on its own it can't determine the exact cause for the danger levels of the health issue, plus the amount of data you get from

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DNA sequencing technology is too low. With AI however, the precision of the data this technology can collect will be high, as once the algorithm or neural network is trained on previous experiments, it can recognize the genomic link to the disease, like pinpointing which kind of mutation and where it is located in the patient's genome. For lymph node metastasis, AI with this DNA sequencing technology controlled by the doctor led to it pinpointing pathological vs non-pathological advancement in the problem. The learning curve and strength was 0.99, better than the diabetic experts. AI algorithms can also standardize variant interpretation, better analyzing the true genetic links to diseases and weeding out the misinformation that can come. Integration with clinical data of different patients is also a massive issue for dna sequencing technology, as even after reading long sequences of dna, and with someone who knows about the human genome, there is still the issue of the relevance that the DNA sequencing technology provides and integrating it with other environmental factors and past health history of different patients. This is crucial for personalized treatments but very difficult to do so as just the primitive qualities of humans is not going to work for every patient especially when it can't be compared to the broader and easier scale of patients that have their data in a biobank. Neural networks in those not broad cases don't work as well. AI based precision medicine platforms, however, can integrate the complex DNA sequencing data with EHRS to allow for properly integrated patient information (Johnson and Wei,2021)[13].

Currently AI based precision medicine is being used for neurological development disorders or NDD's. A challenge for doctors trying to identify the variant and cause of the NDD is figuring out what genes cause the NDD, and how they are being expressed. AI-based precision tech would allow there to be analysis of causal genes, phenotypic heterogeneity, and how the gene is being expressed. With these three components analyzed, doctors would be able to properly identify the variant of the NDD, and properly help the patient get around it while not triggering any of the symptoms. Proper medicine that would not be harmful for the patient would also come out of the help of AI based precision medicine (Uddin,2019)[14].

AI could also help in forensic databases immensely in two different ways. The first way involves using AI to help with the forensic databases privacy problem. Currently, a problem with forensic databases is that the information containing genomic data on people is made public, meaning it has the potential to be manipulated and stolen. With no policy currently in place or education going to let people know of this problem, privacy is a massive issue. AI can help circumvent this issue by first being trained on new data to be able to respond to these new threats. One example of training: The trained machine learning algorithms will be able to detect people trying to steal or manipulate the DNA information. It also will be able to conduct security log analysis, to further help with the privacy issue in forensic databases. Genomic data would become more secure. With AIs increased accuracy on detecting these threats alongside the security log analysis, less money and resources would be spent trying to deal with fake threats (Moisset,2023)[10].

The second way involves AI's usage making it so that law enforcement can be less reliant on genetic samples. With the assistance of forensic databases, AI tools could analyze blood splatter patterns alongside improving the accuracy of toxicology tests. With these two pieces of evidence that can now be looked at with the help of the AI, there will be much faster analysis and organization of DNA samples from crime scenes that are stored in the forensic databases. AI powered tools can analyze so much data at once, so investigators can make fast observations efficiently, so all evidence gets properly checked (Digital Forensics AI: Evaluating, Standardizing and Optimizing Digital,2023)[4].

DNA databases like CODIS are really effective in assisting different law enforcement agencies for solving crimes like homicides and assaults especially, or crimes that are classified to be violent. According to the US department of justice, in 2021, 475000 hits have come, with over 500,000 suspects being identified for around 490,000 investigations. In terms of specific case studies, the Golden State Killer case, using familial DNA within the GED match database to catch the culprit, showcases the power of forensic DNA databases. Fingerprint Databases such as the AFIS are crucial for linking suspects to crime scenes. Eyewitness testimonies, physical evidence and detective work are still vital in solving crimes, but forensic DNA is quite strong.

Limitations

For forensic databases, however, there is still a limitation that exists. Although public awareness campaigns can help increase potential skeptic's' knowledge of genomic science, it may not address the public's distrust of scientific authorities, which runs deeper than just awareness.

There are a few limitations when it comes to AI. AI is able to eliminate a lot of human based errors and it still can't get rid of inaccuracies. AI especially in forensic databases when it has to help analyze so much genetic information, mistakes will occur and there might be data gaps which really might slow down the process of catching criminals or it might accidentally affect/change the information analyzed in the forensic database. For AI-based precision medicine, a data gap or a mistake might result in a slow or worse incorrect prescription for the patient, which is definitely harmful especially if it has to do on the level of looking at causal genes in the genome (Medtech: Digital Health and Wearable Tech Course,2023)[7]. Also, for Brandon Mayfiled when it came to AFIS and fingerprints, he was wrongfully detained for the Madrid Train Bombings because of a mistaken fingerprint match involving forensic database data so mistakes will still happen.

When it comes to assessing a patient's condition AI mod-

els can sometimes be trained on incomplete datasets leading to disparities in health outcomes due to biases in population representation,genetic diversity especially for genomic data when linked with AI. So AI models have to be invested into and trained on diverse and representative datasets to mitigate bias alongside techniques like data augmentation.

Privacy risks and consent issues need to have careful consideration as well in how you handle it, as different risks are present. AI models also have to comply with these separate data privacy laws and act accordingly when using and storing genomic data to protect patient confidentiality. Genomic data sensitivity is also present, where even if the genomic data gets an anonymous marker placed on it, the genomic data can still be pinpointed to identify individuals with the help of other datasets as genomic data is highly individual. Secondary uses of data like data management, where genomic data used for AI-based precision medicine is shared for research purposes raising the question of consent. The solution to no consent, is to set up full transparency of what the data will be used for, content where the patient can decide to have their genomic data stored and researched, and that their preferences can change over time allowing them to update their status on their own genomic data. Data aggregation can happen to a point where using genomic data from multiple sources can lead to security breaches and so the follow up limitation when it comes to AI and privacy risks trying to be solved, is that AI is extremely vulnerable to security risks.

When patient genetic data is so important, the fact that AI can be compromised through cybersecurity attacks which are becoming more dangerous is a really big problem. For forensic databases means that it might not effectively be able to make the information on these forensic databases private, it could just not be effective security at all.

The best thing to do about these security risks is to use ai and carefully encrypt the data in transit and at rest preventing unauthorized access, store data in complaint and secure data centers, and minimize collection of data to whatever is necessary.

Ethically there are also challenges, as modifying a human embryo with human germline editing capabilities along with AI for precision potentially is seen as disrespect to the idea of human DNA heritage. AI and genomics may not be believed as it is just not an acceptable natural idea for many people in society as you are altering something before it comes out of the womb (Coller 2019) [15].

By far however, the biggest limitation for AI is its interpretability. For AI based precision medicine, trust is extremely important for patients and doctors especially when it comes to a disorder that needs long treatment plans. Why a certain treatment and how it works, how is the pathogenicity for the health problem evaluated, and how does the pathogenicity level associates with either just phenotypic symptoms or a certain genetic variant discovered. The last part especially is important as AI is very helpful in figuring out the causes and effects of genetic variants for different disorders, health problems etc. But AI models are currently developing right now and they are hard to interpret for the doctor. If the doctor is not able to figure out how the AI got to its understanding for the recommendation it gives, they won't be able to accurately support the patient and convey information on self-care for the patient.

Case by case rationale is what the medical field has to strive for when using AI, in order to help people, not a general set of cases that AI models give often from being insufficiently trained or just hard to interpret. Visualized model predictions from the AI have to be there as an easier way for doctors to access the knowledge it tries to provide for different patients.

AI Based Precision medicine simply needs more research and time spent in a few areas. Integrating data from multiple layers such as genomics and proteomics to have the complex understanding behind the disease mechanisms and identify therapeutic targets. Analyzing heterogenous genomics data will allow for better disease stratification and treatment selection but not enough research has been done. AI's impact through large scales of clinical trials will have to be done when AI assesses multiple patients and their health to see if it truly has validated its place in healthcare. Human and AI collaboration techniques to help interoperability still need to be developed alongside properly funded programs to make AI work in healthcare across the whole country and lastly ethical implications with AI still need further exploring.

Conclusion

In conclusion, there are many areas where genomics science can be applied, and there is much potential but there needs to be better information on how we as a society can separate proper information that helps us understand the potential of genomics and not false information that sways bias into medical, law and societal decisions.

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