Abstract

In recent years, Whole Genome Sequencing (WGS) evolved from a futuristic-sounding research project to an increasingly affordable technology for determining complete genome sequences of complex organisms, including humans. This prompts a wide range of revolutionary applications, as WGS promises to improve modern healthcare and provide a better understanding of the human genome – in particular, its relation to diseases and response to treatments. However, this progress raises worrisome privacy and ethical issues, since, besides uniquely identifying its owner, the genome contains a treasure trove of highly personal and sensitive information. In this article, after summarizing recent advances in genomics, we discuss some important privacy issues associated with human genomic information and identify a number of particularly relevant research challenges.

1 Introduction

Recent years have witnessed impressive advances in DNA sequencing. Both throughput and affordability of new-generation sequencing platforms have increased at a pace faster than Moore’s Law would otherwise predict. It seems quite reasonable to assume that, in a few years, most individuals in developed countries will have the means of having their genomes sequenced, thus enabling personalized genomic medicine and facilitating preventive treatment and diagnosis.

However, for now this remains only a prospect and much more research is needed to understand the very complex relationship between genome and health. To conduct this research, the scientific community needs large cohorts of patients (or volunteers) willing to share their genetic material. For instance, the Personal Genome Project involves participants that agree to have their genomic data – along with other personal information – made publicly available on the Internet, which raises many potential privacy, ethical, and legal concerns.

The first documented case of privacy issues dates back to the end of the 19th century, triggered by the availability of a new and revolutionary observation and identification tool: the photo camera. Since then, several other such tools have become widespread, including: video cameras, credit cards, Web browsers, and mobile phones. These tools reveal our presence and habits in various spheres of life, as well as our communication and mobility patterns. DNA sequencing greatly exacerbates this problem, as the genome represents our ultimate biological identity. By combining genomic data with information about one’s environment or lifestyle (often easily obtainable from social networks), could make it possible to infer that individual’s phenotype.

In general, access to genomic data prompts some important privacy concerns: (i) DNA reflects information about genetic conditions and predispositions to specific diseases such as Alzheimer’s, cancer, or schizophrenia, (ii) DNA contains information about ancestors, siblings, and progeny, (iii) DNA (almost) does not change over time, hence revoking or replacing it (as with other forms of identification) is impossible, and (iv) DNA analysis is already being used both in law enforcement and healthcare, thus prompting numerous ethical issues. Furthermore, it is hard to assess or estimate the extent of the personal information that could be extracted or derived from the genome in the future.

In this article, after briefly over-viewing some basic
genomic concepts, we describe some expected benefits of personalized medicine and discuss notable privacy issues, as well as associated research challenges.

2 Background

This section provides a brief genomics primer.

2.1 Processing chain

The human genome is encoded in double stranded DNA molecules consisting of two complementary polymer chains. Each chain consists of simple units called nucleotides (A,C,G,T). The DNA of a person can be retrieved from various sources (e.g., saliva, hair, skin, blood). Once a sample is collected, the genetic material is extracted and then sequenced – using a DNA sequencing platform – to obtain the so-called raw DNA sequence. This is usually in the form of short reads, each including hundreds of nucleotides from random parts of the genome. Next, the raw reads are quality-controlled, analyzed, and aligned to the reference genome (digital nucleic acid sequence database, assembled by scientists as a representative example of our species’ set of genes), allowing the progressive reconstruction of the whole sequenced genome.

The collection of all aligned raw reads is usually a SAM (sequence alignment/map) file. There are hundreds of millions of short reads (each including around 100 nucleotides) in the SAM file. Each nucleotide is included in several short reads to have high coverage of each subject’s DNA. After further analysis of the SAM file, eventually, the approximately 3.2 billion letters in the DNA sequence of the person are reconstructed.

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2.2 Genetic Variations

Even though most of the DNA sequence is conserved across the whole human population, around 0.5% of each person’s DNA (which corresponds to several millions of nucleotides) is different from the reference genome, owing to genetic variations. Single nucleotide polymorphism (SNP) is the most common DNA variation. A SNP is a position in the genome holding a nucleotide that varies between individuals. For example, in Fig. 1, two sequenced DNA fragments from two individuals contain a single different nucleotide at a particular SNP position (i.e., locus). Multiple Genome-Wide Association Studies (GWAS) performed in recent years have shown that a patient’s susceptibility to particular diseases can be partially predicted from sets of his SNPs [16, 33]. For example, it was reported that there are three genes bearing a total of ten particular SNPs necessary to (partially) analyze susceptibility to Alzheimer’s disease [46]. Thus, leakage of SNPs often poses a significant threat to individual privacy. Each SNP contributes to the susceptibility in a different amount and the contribution amount of each SNP is determined by Genome-Wide Association Stud-
ies (GWAS) [54] on case and control groups (these studies are published in several papers).

Each SNP position includes two alleles (i.e., two nucleotides) and everyone inherits one allele of every SNP position from each of his parents. If an individual receives the same allele from both parents, he is said to be homozygous for that SNP position. If, however, he inherits a different allele from each parent (one minor and one major), he is called heterozygous. It is important to note that a SNP becomes a variant when it carries at least one minor allele.

There are approximately 40 million approved SNPs in the human population as of now (according to the NCBI dbSNP [40]) and each patient carries on average 4 million variants (i.e., SNPs carrying at least one minor allele) out of this 40 million. We note that the number of approved SNPs in human population is increasing very rapidly [40], whereas the number of variants per patient (around 4 million) remains the same. Moreover, this set of 4 million variants is different for each patient.

### 2.3 Linkage Disequilibrium

An interesting characteristic of the SNPs, called Linkage Disequilibrium (LD) [17], poses a notable privacy threat. LD is observed whenever SNPs are not independent of each other. Therefore, the nucleotide of a certain SNP can be inferred from the contents of other SNP positions using the LD relationship. The most well-known example of the aforementioned threat is the ApoE status of Jim Watson (the co-discoverer of DNA), who published his genome with the exception of his ApoE gene (which carries SNPs to determine the risk for Alzheimer’s disease). However, it was later shown that these SNPs on his ApoE gene can be (probabilistically) inferred using their LD relationships with the published ones [43]. For example, assume that SNP \(_i\) and SNP \(_j\) (SNPs which reside at positions \(i\) and \(j\) on the DNA sequence, respectively) are in LD. Let \((A_1, A_2)\) and \((B_1, B_2)\) be the potential alleles for these two SNP positions (i.e., loci) \(i\) and \(j\). Further, let \((p_1, p_2)\) and \((q_1, q_2)\) be the allele probabilities of \((A_1, A_2)\) and \((B_1, B_2)\), respectively. That is, the probability that an individual will have \(A_1\) as the first allele of SNP \(_i\) is \(p_1\), and so on. (Recall that each SNP position includes two alleles, i.e., two nucleotides.) If there were no LD (i.e., if SNP \(_i\) and SNP \(_j\) were independent), the probability that an individual will have both \(A_1\) and \(B_1\) as the first alleles of SNP \(_i\) and SNP \(_j\) would be \(p_1q_1\). However, due to the LD, this probability is equal to \(p_1q_1 + D\), where \(D\) represents the LD between these two SNP positions. In Fig. 2, we illustrate this LD relationship for all possible combinations of \((A_1, A_2)\) and \((B_1, B_2)\).

### 3 Towards Personalized Medicine

Widespread and affordable availability of fully sequenced human genomes creates enormous opportunities, which we summarize in Fig. 3 (and discuss in this section).

In particular, whole genome sequencing (WGS) facilitates the advent of a new era of predictive, preventive, participatory, and personalized medicine (“P4 medicine”) [31]. Personalized genomic medicine is recognized as a significant paradigm shift and a major trend in health care [58], where treatment and medication type/dosage would be tailored to the precise genetic makeup of individual patients.

For instance, certain genetic mutations are known to alter drug metabolism, thus genomic tests are often used today to predict a patient’s response to particular drugs. The study of the impact of genetic variations on the response to medications is called pharmacogenomics. A well-known example in this direction includes testing for SNP mutations in the \(tpmt\) gene for childhood leukemia patients, prior to prescribing 6-Mercaptopurine and Azathioprine drugs. The \(tpmt\) gene codes for the TPMT enzyme that metabolizes these drugs. Moreover, genetic polymorphisms affecting enzymatic activity of TPMT are correlated with variations in sensitivity and toxicity response to such drugs. Other common examples include pre-testing for Zelboraf (Roche’s treatment for advanced skin cancer), as well as pre-treatment testing for Philadelphia
chromosome mutations related to Acute Lymphoblastic Leukemia (ALL) and BRCA1/BRCA2 genes in correlation to familial breast and ovarian cancer syndromes. Experts estimate that about a third of the 900 cancer drugs currently in clinical trials could soon come to market with an enclosed recommendation for a DNA or another molecular test [10].

Vanderbilt University’s PREDICT program (Pharmacogenomic Resource for Enhanced Decisions in Care and Treatment) [38] helps physicians tell which drugs are most likely to work for their patients, and which they should avoid, based on the genetic characteristics of the patients, instead of long trial and error periods. For instance, [59] reports how a specific cholesterol-lowering drug was successfully selected based on the genomic profile of a patient with coronary artery disease.

Experts predict that advances in WGS will further stimulate advances in personalized medicine [23]. Commercial entities, such as Knome, already offer services that take raw genomic data and create usable reports for doctors. In general, availability of a patient’s fully sequenced genome will enable clinicians, doctors, and testing facilities to run a number of complex and correlated genetic tests in a matter of seconds, using specialized computational algorithms, as opposed to more expensive and slower in vitro tests.

Another recent Canadian study has shown how, for some cardiac patients, recovery from a common heart procedure can be complicated by a single gene responsible for drug processing, and that selection of blood thinner drugs should depend on whether or not patient holds such a gene mutation [60]. Cancer treatment is also one of the most predominant application fields of personalized medicine.

The democratization of low-cost whole genome genotyping and sequencing provides individuals with direct access to their genomic information, including to some genetic disease risk tests. For example, a well-known commercial entity, 23andMe [1], provides relatively low-cost genetic ancestry and disease risk tests for 960,000 specific SNPs. However, 23andMe does not yet offer WGS. Fig. 4 illustrates genetic disease risk results of a real human with a fictional name “Greg Mendel” provided by 23andMe. It shows the diseases for which Greg Mendel’s calculated risk is higher than average. In [52], Topol mentions a few real stories about how the disease risk values obtained from 23andMe helped early diagnosis of serious diseases.

While personalized medicine creates a lot of research “enthusiasm”, a number of biomedical experts have also expressed doubts related to the limits of gene mapping’s power to predict a person’s likelihood of developing a disease [39]. This is because it is not always clear to
which extent certain diseases are correlated to genetic or environmental factors (a list of diseases known to be associated to genetic features is available in [16]).

Availability of WGS will also facilitate faster and lower-cost digital versions of genetic tests that are currently performed in vitro. For instance, computational paternity testing can be designed to mimic its in vitro counterpart, with greater speed and accuracy, while preserving its legal acceptance. Furthermore, ancestry and genealogical testing is already offered by several commercial entities. In such tests, publicly available genomic data from individuals belonging to different ethnic groups is compared against the customers’ genomic information to understand how the customers relate to known ethnic groups. Similarly, genetic compatibility tests, which let potential or existing partners assess the risk of transmitting to their children genetic diseases with Mendelian inheritance [37], are offered by various online services.

Genomic privacy is often viewed with skepticism, since every individual constantly sheds – or otherwise leaves behind – his biological “fingerprints,” such as hair, skin, or saliva. This material can be collected (even much later) and used for DNA sequencing. However, such attacks pose a credible threat only against a targeted individual or a small group of people. The danger is clearly incomparable with privacy threats posed by access to large numbers of digitized genomes – the main focus of this paper.

Furthermore, traditional approaches to privacy, such as de-identification or aggregation [35], are ineffective in the genomic context, since the genome itself is the ultimate identifier [29]. For instance, a recent study by Gymrek et al. [25] demonstrated feasibility of re-identifying DNA donors from a public research database using information available from popular genealogy Web sites and other available information. Additional work on genomic re-identification includes [45] and [29].

Moreover, the range of possible abuses is broadened by the increasingly common handling and sharing of health information (in electronic form) among insurance companies, health care providers and employers. Unfortunately, keeping digital records secure is a problem [47]. For instance, medical information of 34,000 patients was leaked from Howard University Hospital; also, hackers compromised servers of Utah Department of Health and stolen medical information of almost 800,000 individuals.

The privacy problem is further exacerbated by the
fact that genomes of any two closely related individuals are highly similar. Thus, disclosure of one’s genome leads to leakage of significant genomic information about that person’s close relatives, including parents, siblings and offspring. This is a problem regardless of how the disclosure occurs: voluntarily, accidentally or maliciously. Therefore, genomic privacy is a unique issue, since, in most other privacy-sensitive scenarios, only the individual’s data is at stake. Whereas, in the genomic context, disclosure of personal information impacts a potentially large group of individuals. The most recent example of this issue is the controversy between family members of the deceased Henrietta Lacks (whose genome was sequenced and published after her death, without getting the permission of her family) and scientists who are in favor of publishing genomes online for research purposes [50].

Even more worrying is that consequences of genomic data disclosure are not limited in time. In certain privacy leakage scenarios, some recourse is possible. For example, bank account numbers and passwords can be changed, physical or electronic documents (even public key certificates) can be replaced and old ones can be revoked. In contrast, a genome is neither mutable nor “revokable”. Moreover, as large portions thereof are passed on to future generations, disclosure of one’s genomic information can turn into an endless curse for future generations.

Based on the above, it is not surprising that privacy concerns represent a formidable obstacle for assembling large human genomic databases, e.g., for the purpose of conducting Genome-Wide Association Studies (GWAS). More generally, privacy concerns might actually stand in the way of advances in medicine and consequent improvements in overall healthcare. The same could apply in the domain of law enforcement where DNA-based identification is being increasingly used and there is a need for secure and reliable handling of large numbers of genomes.

In US, the federal government has been aware of privacy and ethical issues in genomics. For example, as early as 1990, the National Human Genome Research
Institute (NHGRI) established the Ethical, Legal and Social Implications (ELSI) Research Program with the goal of exploring repercussions of advances in genetic and genomic research on individuals, families and communities.

Federal laws, such as the 2003 Health Insurance Portability and Accountability Act (HIPAA), provide a general framework for protecting and sharing Protected Health Information (PHI). Furthermore, the Genetic Information Nondiscrimination Act (GINA) adopted in 1998 prohibits discrimination on the basis of genetic information with respect to health insurance and employment [55]. Also, some states, e.g., California, have recently started to consider DNA privacy laws [48].

Even the popular culture, via sci-fi movies and literature, has touched upon genetic discrimination. For instance, the notion of genism that originated in the 1997 movie “GATTACA”, refers to the theory that distinctive human characteristics and abilities are determined by genes, resulting in discrimination as pernicious as racism [2]. Influenced by this movie, a prominent molecular biologist wrote: “Gattaca is a film that all geneticists should see if for no other reason than to understand the perception of our trade held by so many of the public-at-large” [49].

While providing general guidelines, current legislation does not offer sufficient technical information about safe and secure ways of storing and processing digitized genomes. We believe that security and privacy issues for genomic data (in the context of both individual genomes and databases thereof) are timely, important and relatively poorly understood.

Privacy practitioners and consumer organizations are strongly advocating the need for more restrictive legislation as a result of gaps in current policies. Also, the Electronic Frontier Foundation (EFF) has analyzed warrant-less DNA gathering from suspects and against DHS’s efforts to collect genetic data from people placed into administrative detention. A recent report from the US Presidential Commission for the Study of Bioethical Issues [44] analyzed advances of WGS, and highlighted growing privacy and security concerns. This report makes a few privacy and security recommendations, including, unfortunately, de-identification.

At the policy level, challenges include the need for informed consent to guard against surreptitious DNA testing. Authorities and companies should obtain written permission from citizens before collecting, analyzing, storing or sharing their genetic information, e.g., preventing collection of hair or saliva samples and using them for unauthorized sequencing.

On the other hand, some academics fear that restrictive (privacy-friendly) measures could seriously hinder genomic research. Scientists typically sequence DNA from large numbers of people in order to determine genes associated with particular diseases. The informed consent restriction would mean that large genomic datasets could not be re-used to study a different disease; researchers would either need to destroy the data after each study, or track down all previously enrolled study participants for each new authorization. Also, similarity of related individuals’ genomes raises doubts as to whether relatives should also provide consent.

Finally, collection and analysis of human genomes does not arise only in the contexts of research studies and improved healthcare. It also comes up in increasingly popular commercial (for-profit) applications, which are not well-regulated. An example is genepartner.com, which claims to do matchmaking based on unclear genetic features.

5 Existing Work on Genomic Privacy

Due to the sensitivity of genomic data, research on the privacy of genomic data has accelerated over the past few years. First, a few techniques have been proposed for secure and privacy-preserving computation on DNA fragments/snippets. More recently, the security community started focusing on fully-sequenced genomes, motivated by the advances in WGS.

In [53], Troncoso-Pastoriza et al. propose a protocol for string searching (then re-visited by Blanton and Aligasari [8]), where one party with his own DNA snippet can verify the existence of a short template within his snippet by using a Finite State Machine (FSM) in an oblivious manner. Also, secure pattern matching techniques, e.g., those in [22] and [28], have been applied to securely search binary strings in a DNA snippet. Katz et al. [34] realize secure computation of the CODIS test [51] (run by the FBI for DNA identity testing) and other search tests that could not be otherwise implemented using pattern matching or FSM.

To compute the similarity of DNA sequences, in [32], Jha et al. propose techniques for privately computing the edit distance of two strings by using garbled circuits.
In [9], Bruekers et al. propose privacy-enhanced comparison of DNA profiles for identity and paternity tests, using homomorphic encryption on DNA snippets.

Baldi et al. [6] are the first to focus on whole genomes and introduce several cryptographic protocols, based on Private Set Operations, that realize secure testing of whole human genomes, e.g., paternity tests and genetic screening for personalized medicine or recessive genetic diseases. In their setting, individuals obtain their genomes and allow authorized parties (e.g., doctors and clinicians) to run genetic tests such that only test results are disclosed to one or both parties (with provable security). In a follow-up work, De Cristofaro et al. propose a framework and implement a toolkit, called Genodroid [15] for privacy-preserving genomic tests on Android smartphones. In [11], Canim et al. propose securing biomedical data using cryptographic hardware.

Ayday et al. [3, 4, 5] also focus on the privacy of personal use of genomic data (e.g., in medical tests and personalized medicine methods), and propose methods for protecting user’s genomic privacy by considering the statistical relationship between the variants.

When releasing databases consisting of aggregate genomic data (e.g., for research purposes), known privacy-preserving approaches, such as de-identification, are ineffective on (un-encrypted) genomic data [56, 36]. Homer et al. [30] prove that the presence of an individual in a case group can be determined by using aggregate allele frequencies and his DNA profile. In another study [24], Gitschier shows that a combination of information, from genealogical registries and a haplotype analysis of the Y chromosome collected for The HapMap Project, allows for the prediction of the surnames of a number of individuals held in the HapMap database. Thus, releasing (aggregate) genomic data is currently banned by many institutions due to this privacy risk. In [61], Zhou et al. study the privacy risks of releasing the aggregate genomic data. They propose a risk-scale system to classify aggregate data and a guide for the release of such data. Recently, Fienberg et al. [18] use differential privacy to ensure that two aggregated genomic databases, differing from each other by only one individual’s data, have indistinguishable statistical features. However, this method does not work well for sparse databases and severely affects the reliability of the genomic data (due to noise injection).

Finally, Wang et al. [57] propose a privacy-protection framework for important classes of genomic computations (e.g., search for homologous genes), in which they partition a genomic computation, distributing sensitive data to the data provider and the public data to the data user. Also, Chen et al. [14] propose a secure cloud-based algorithm to align short DNA sequences to a reference (human) DNA sequence (i.e., read mapping).

6 Open Research Problems

As discussed above, advances in genomics will soon result in large numbers of individuals having their genomes sequenced and obtaining digitized versions thereof. This poses a wide range of technical problems, which we explore below.

Storage and Accessibility: Genome at Rest. Due to its sensitivity and size (about 3.2 billion nucleotides), one key challenge is where and how a digitized genome should be stored. It is reasonable to assume that an individual who requests (and likely pays for) genome sequencing should store the result, as is already the case with any other personal medical results and information. This raises numerous issues, including:

1. Should the genome be stored on one’s personal devices, e.g., a PC or a smartphone? If so, what, if any, special hardware security features (e.g., tamper-resistance) are needed?
2. Can it be outsourced to a cloud provider?
3. Should the sequencing facility keep an escrowed copy of the genome?
4. Should it be entrusted to one’s personal physician and/or health insurance provider?
5. How is it to be stored: in the clear or encrypted? If the latter, where are encryption keys generated: at the lab? at owner’s premises? at the cloud provider? Where are these keys stored?
6. How to guarantee integrity and authenticity of the digitized genome?
7. Should backups be made? If so, how often and where can copies be kept?
8. How can one erase a genome securely?
9. Should an individual periodically re-sequence their genome to take advantage of more accurate technology?

Privacy: Genome in Action. Given the genome’s sensitivity, an individual should, ideally, never disclose any
information contained therein. However, this would prevent the access to any genomic application that cannot be entirely and securely performed in situ, i.e., within a secure perimeter of one’s own personal device. In principle, this might be possible if operations are performed in some standardized and certified form. For example, if testing for a genetic disease requires matching a well-known pattern in some approximate location in the genome, that pattern and its parameters can be certified by some trusted agency (such as the US Food and Drug Administration). Thus, an individual could be assured that a legitimate test for a specific genetic disease is being conducted and the result is clearly communicated to that individual; the latter would then have the option to keep the result private.

At the same time, it is hard to foresee the range and complexity of future genetic operations: some (future) tests might be too computationally complex to be performed within the confines of a personal device. Furthermore, some genetic testing would probably involve multiple genomes, e.g., when tracing origins of some conditions, siblings or parents/children might need to be tested together. Similarly, in assessing risks of genetic conditions for future progeny, both prospective parents have to be tested. Also, some genetic tests constitute intellectual property of a pharmaceutical/biomedical company (which needs to be protected) [41, 27, 13].

As soon as genomic information leaves the (virtual) hands of its owner, purely technical approaches to privacy become insufficient. Legal and professional guidelines are certainly needed to govern how information is transmitted, stored, processed, and eventually disposed of on the receiving end, e.g., by the physician, hospital, pharmacist or medical lab.

Long-term data protection. Even if genomes are encrypted, encryption schemes considered strong today might gradually weaken in the long term, whereas genome sensitivity does not dissipate over time. It is not too far-fetched to imagine that a third-party in possession of an encrypted genome might be able to decrypt it years or decades later. For instance, the Advanced Encryption Standard (AES) scheme supports key lengths up to 256 bits – a key length estimated by NIST, following Moore’s law, to be secure several years after 2030 [42]. However, computational breakthroughs or unforeseen weaknesses might allow breaking the encryption earlier than expected. Also, even leakage of a long-deceased individual’s genome could affect genomic privacy of that person’s living progeny.

Assuming that it can not be copied, an encrypted genome could be periodically re-encrypted. Alternatively, one could split the genome, using secret-sharing techniques, and partition it among several providers. However, this opens the problem of efficient reassembly of the genome for various operations as well as how to guarantee non-collusion between providers.

Accuracy and Accountability: Computational genomic tests should guarantee accuracy at least equivalent to that of their current analog in vitro counterparts. For example, a software implementation of the paternity test should offer at least the same confidence as its in vitro counterpart currently admissible in a court of law. Also, computational tests should aim at accountability, e.g., by providing lasting guarantees of correctness for both execution and input information.

Efficiency. Computational genomic tests should incur minimal communication/computational costs. Minimality in this setting is relative to the context of such tests. For instance, patients may be inclined (and accustomed) to wait several days to obtain results of genetic tests that concern their health. However in the computational setting, long running times on personal devices might hinder the real-world practicality of these tests (besides negating one of the main motivations for computational tests.)

Usability. Computational genomic tests that involve end-users should be usable by, and meaningful to, regular non-tech-savvy individuals. This translates into non-trivial questions, such as: how much understanding should be expected from a user running a test? What information (and at what level of granularity) should be presented to the user as part of a test and as its outcome? Do privacy perceptions and concerns experienced by patients match those expected by the scientific community? Some users might be willing to forego their genomic privacy in some certain cases. For instance, one may think that patients will be likely to reveal their genomes to their medical doctors (and hence trade off privacy of their genomes) to enable tests that can save them from, e.g., cancer. In contrast, in the case of online services or pharmaceuticals, an individual might not wish to forgo privacy. However, very few efforts
(e.g., [20]) has focused on users’ concerns, thus prompting the need for ethnographic studies. Also, there remains an open problem of how to effectively communicate to the users potential privacy risks associated with genomic information and its disclosure.

**Large-scale research on human genomes.** As discussed in Section 4, potential privacy, legal, and ethical concerns appear to conflict with large-scale research on human genomes, such as Genome-Wide Association Studies (GWAS). However, large scale studies are needed to discover associations between genetic make-up and medical conditions. One current trend is to store donors’ genomes in the cloud and use analytics techniques running on powerful computer clusters. Once again, this prompts many privacy and legal concerns.

7 Conclusion

This paper discussed some “chills and thrills” of an emerging phenomenon – affordable and readily available genomic sequencing. As something radically novel, it brings great opportunities and significant concerns, especially pertaining to personal privacy. Mitigating privacy issues will require long-term collaboration among geneticists, other healthcare providers, ethicists, lawmakers, and computer scientists. As one of the first steps towards such a collaboration, we are involved in organizing a multi-disciplinary seminar on genomic privacy [26]. In order to foster this collaboration, funding agencies need to target this topic. Until recently, at least in the United States, genomic privacy unfortunately fell into a sort of a “funding gap” between several agencies. One obvious candidate for playing a key funding role is the National Institute of Health (NIH). Yet, although it covers both bioinformatics and WGS ethical issues, NIH has funded little research in the genomic privacy context. The National Science Foundation (NSF), the main agency responsible for funding academic computer-science research, recently initiated a “Smart and Connected Health” program that includes so-called “integrative projects” requiring collaboration among computer and health sciences. It remains to be seen whether this program will engender long-range genomic privacy research. Other US funding agencies have not, thus far, targeted genomic privacy. A similar situation can be observed in Europe: of course, there are numerous EU and nationally funded projects focusing on e-health, some of which address data protection.

However, the genomic privacy challenge has been overlooked, and the number of computer scientists working on the topic is even lower than in the United States. An additional issue is that, although most officials in charge of data protection typically have a strong legal background, they lack computer science expertise. Consequently and not surprisingly, they tend to rely on legislation more than on technology.

In conclusion, we hope that the privacy issues highlighted in the article will be addressed promptly and encourage collaboration among researchers in the fields outlined above. We believe that consideration of such privacy issues will have a positive benefit to society and individuals in their daily lives.

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