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THE HUMAN GENOME AS PUBLIC: JUSTIFICATIONS AND IMPLICATIONS

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ABSTRACT

Since the human genome was decoded, great emphasis has been placed on the unique, personal nature of the genome, along with the benefits that personalized medicine can bring to individuals and the importance of safeguarding genetic privacy. As a result, an equally important aspect of the human genome – its common nature – has been underappreciated and underrepresented in the ethics literature and policy dialogue surrounding genetics and genomics. This article will argue that, just as the personal nature of the genome has been used to reinforce individual rights and justify important privacy protections, so too the common nature of the genome can be employed to support protections of the genome at a population level and policies designed to promote the public's wellbeing.

In order for public health officials to have the authority to develop genetics policies for the sake of the public good, the genome must have not only a common, but also a public, dimension. This article contends that DNA carries a public dimension through the use of two conceptual frameworks: the common heritage (CH) framework and the common resource (CR) framework. Both frameworks establish a public interest in the human genome, but the CH framework can be used to justify policies aimed at preserving and protecting the genome, while the CR framework can be employed to justify policies for utilizing the genome for the public benefit. A variety of possible policy implications are discussed, with special attention paid to the use of large-scale genomics databases for public health research.

INTRODUCTION

Since the human genome was decoded at the turn of the millennium, health advocates, medical researchers and policymakers have stressed the unique, personal nature of the genome. Correspondingly, the benefits that personalized medicine can bring to individuals and the importance of safeguarding genetic privacy have also received much attention.[1] Although the claim that DNA is uniquely individuating is incontrovertible (indeed, even identical twins do not have identical genomes),[2] the focus on the personal character of the human genome has siphoned attention away from an equally important aspect of the genome: its common nature. The human genome is what distinguishes human beings from other life forms, allows us to procreate, and is the literal and figurative heritage of humankind. The focus on the personal character of the genome has had a skewing impact on the policy dialogue surrounding genetics and genomics; policy discussions and developments have concentrated on how to protect and utilize a person's unique genetics at the expense of advancing policies that

[1] National Institutes of Health Precision Medicine Initiative. About the Precision Medicine Initiative Cohort Program. Available at: https://www.nih.gov/precision-medicine-initiative-cohort-program [Accessed 14 Apr 2016].

[2] C.E. Bruder et al. Phenotypically concordant and discordant monozygotic twins display different DNA copy-number-variation profiles. *Am J Hum Genet* 2008; 82(3): 763–771.

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maximize our ability to use genomics for the public good. This article will argue that, just as the personal nature of the genome has been used to reinforce individual rights and justify important privacy protections, the common nature of the genome can be employed to support protections of the genome at population level and policies designed to promote the public's wellbeing.

In order to explore the kinds of protections and policies that can be grounded in the common aspect of the human genome, we must carefully examine what we mean by 'common' and 'public' in the DNA context. This article will argue that DNA carries a public dimension through the use of two conceptual frameworks: the common heritage (CH) framework and the common resource (CR) framework. These frameworks have been developed by other genomic theorists,[3] but will be examined further in this article and applied in a novel way – to establish the public character of the genome and consider the resulting authority of public health officials to create policies on genetics that promote the public good. Both frameworks establish a public interest in the human genome, but the CH framework can be used to justify policies aimed at preserving and protecting the genome, while the CR framework can be employed to justify policies for utilizing the genome for the public benefit. While a variety of possible policy implications will be discussed, special attention will be paid to the use of large-scale genomics databases for public health research. It should be noted that the goal of this article is *not* to devalue the view that the genome is personal and that genetic privacy ought to be respected. Rather, it aims to explore the policy implications that result from more fully appreciating the common and public components of the genome, balancing respect for individual autonomy with pursuit of the public interest.

THE GENOME AS PERSONAL AND PRIVATE

DNA is personal in a fundamental sense; it exists physically within the person. The physical nature of DNA has played an important role in public and legal discourse regarding the status of the human genome.[4] For instance, in the gene patenting debate leading up to the United States Supreme Court's *Myriad Genetics* decision in 2013, some argued that 'The government should not be granting private entities control over something as personal and basic to the human body as our genes.'[5] However, despite the physical presence of DNA within our bodies, the US Supreme Court did not consider police seizure of DNA by cheek swabbing to be an unfair violation of personal privacy in *Maryland v. King* (2013). The court held that 'taking and analyzing a cheek swab of the arrestee's DNA is, like fingerprinting and photographing, a legitimate police booking procedure that is reasonable under the Fourth Amendment.'[6] *Maryland v. King* pertained to the authority of law enforcement personnel to access DNA, which is analogous but not identical to the potential authority of public health officials to access people's genomic information. Thus while the court's consideration of DNA's presence within our bodies reaffirms the personal, physical nature of the genome, the court's ultimate rejection of the personal privacy argument in this case does not have clear implications for the public health context.

Perhaps more important than the physical nature of DNA is the fact that the genome is unique to particular people. The individualistic nature of the genome is the basis of personalized medicine,[7] which aims to tailor treatments to individuals' specific genetic characteristics.[8] The promise of personalized medicine is a potential benefit of the personal aspect of the human genome. One drawback of the unique nature of the genome, however, is that an individual's DNA is always identifiable. An individual's genome can be identified even when it is aggregated with 1,000 other samples.[9] Given that DNA cannot reliably be de-identified, it is understandable for people to have strong concerns about genetic privacy.

A final sense in which DNA is personal is in the intimate nature of the information it contains. The genome contains sensitive information about people and those closest to them, mediating relationships between family members.[10] Furthermore, some view the information held in DNA as essential to an individual's identity.[11] Regardless of one's views on genetic essentialism, it is clear that the information contained in DNA is at least as intimate and sensitive as other information contained

[3] See, e.g. M. Queloz. The Double Nature of DNA: Reevaluating the Common Heritage Idea. *J Polit Philos* 2016; 24(1): 47–66; P.N. Ossorio. The human genome as common heritage: common sense or legal nonsense? *J Law Med Ethics* 2007; 35(3): 425–439.

[4] M. Queloz, *op. cit*. note 3.

[5] American Civil Liberties Union. The Fight to Take Back Our Genes. Available at: https://www.aclu.org/feature/fight-take-back-our-genes [Accessed 14 Apr 2016].

[6] Maryland v. King, 569 US __ (2013).

[7] G. D. Smith et al. Genetic epidemiology and public health: hope, hype, and future prospects. *Lancet* 2005; 366(9495): 1484–1498.

[8] The White House Statements and Releases. 30 Jan 2015. Fact Sheet: President Obama's Precision Medicine Initiative. Available at: https://www.whitehouse.gov/the-press-office/2015/01/30/fact-sheet-president-obama-s-precision-medicine-initiative [Accessed 14 Apr 2016].

[9] A.L. McGuire. Identifiability of DNA data: the need for consistent federal policy. *Am J Bioeth* 2008; 8(10): 75–76; N. Homer et al. Resolving individuals contributing trace amounts of DNA to highly complex mixtures using high-density SNP genotyping microarrays. *PLoS Genet* 2008; 4(8): e1000167.

[10] S. Dheensa, A. Fenwick & A. Lucassen. 'Is this knowledge mine and nobody else's? I don't feel that.' Patient views about consent, confidentiality and information-sharing in genetic medicine. *J Med Ethics* 2016; 42(3): 174–179.

[11] C. Hauskeller, S. Sturdy & R. Tutton. Genetics and the Sociology of Identity. *Sociology* 2013; 47(5): 875–886.

in a person's medical record, including family history of disease. Arguably, genetic information is even more sensitive because of its predictive nature,[12] although some overestimate and overemphasize the deterministic quality of DNA.[13]

The physical nature of DNA, its uniqueness to each individual, and the intimate information it contains, all contribute to the conception of the genome as personal. Because of the personal nature of the genome, genetic privacy has become a central value in society's response to the increased prevalence of genetic information. Peeking into or making use of genetic information without an individual's permission is considered a serious invasion of privacy,[14] one which can even 'disrupt our very sense of self'.[15] The intrinsic value of genetic privacy is based on the principle of autonomy and individual self-governance; people should be able to control who has access to such personal information.[16]

Public opinion about possible uses of genetic data confirm that many people are concerned about potential violations of genetic privacy. Etchegary et al. found that participants across all sections of their Canadian study on the perceptions and expectations of genetics research had concerns about genetic privacy, including who would have access to their data and how it would be protected.[17] In the US, Kerath et al. found that respondents to their survey on participation in genetics research were most concerned about the privacy of their medical and genetic information.[18] Similarly in Europe, public support for biobanks was shown to be lower for those with high levels of concern about privacy, and privacy concerns were frequently mentioned in focus groups on the topic.[19] Members of the public take genetic privacy seriously, which explains why genetic privacy has been so dominant in the ethics literature on genetics and genomics research to date.

To reflect the importance of genetic privacy to many individuals, international and national legal instruments have been developed to ensure that genetic privacy is not violated. UNESCO's Universal Declaration on the Human Genome and Human Rights states that 'Genetic data associated with an identifiable person and stored or processed for the purposes of research or any other purpose must be held confidential in the conditions set by law' and that 'no one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.'[20] Similarly, the US Genetic Information Nondiscrimination Act of 2008 was designed to prevent discrimination in health insurance and employment on the basis of our genes by limiting insurer and employer access to genetic information, including family medical history. While the Act does not refer explicitly to the concept of genetic privacy, implicit in its premise – that employers and health insurance companies are not entitled to genetic information that is traceable to individuals – is the notion that genetic privacy ought to be respected. Furthermore, the 2015 Notice of Proposed Rulemaking (NPRM) for revising the Common Rule (federal guidelines for human subjects research) contains separate sections devoted to ensuring genetic privacy. For instance, one option for the final Common Rule proposes to expand the definition of 'human subjects' to any biospecimen that is potentially identifiable, including specimens that contain 'even small portions of a person's genome'.[21] While some have criticized the NRPM's stringent approach to biospecimen regulation,[22] the NPRM is an initial attempt to respond to public concerns about the privacy of potentially identifiable specimens, particularly given apprehension about the inability to reliably de-identify samples containing genetic material.[23] A thorough discussion of whether there is a strong basis for treating genetic material differently from other kinds of medical information is beyond the scope of this article. However, it is reasonable to assume that the information contained in the genome is extremely personal, which explains why DNA is subject to privacy provisions that are at least as protective as those aimed at securing other medical information.

[12] P. Brodwin. Genetics, identity, and the anthropology of essentialism. *Anthropol Quart* 2002; 75(2): 323–330.
[13] R.C. Green, D. Lautenbach & A.L. McGuire. GINA, genetic discrimination, and genomic medicine. *N Engl J Med* 2015; 372(5): 397–399.
[14] A. Gutmann. Data re-identification: prioritize privacy. *Science* 2013; 339(6123): 1032.
[15] P.A. Roche & G.J. Annas. DNA testing, banking, and genetic privacy. *N Engl J Med* 2006; 355(6): 545–546.
[16] L. Jamal et al. Research participants' attitudes towards the confidentiality of genomic sequence information. *Eur J Hum Genet* 2014; 22(8): 964–968.
[17] H. Etchegary et al. Community engagement with genetics: public perceptions and expectations about genetics research. *Health Expect* 2015; 18(5): 1413–1425.
[18] S.M. Kerath, G. Klein & M. Kern. Beliefs and attitudes towards participating in genetic research - a population based cross-sectional study. *BMC Public Health* 2013; 13: 114.
[19] G. Gaskell et al. Publics and biobanks: Pan-European diversity and the challenge of responsible innovation. *Eur J Hum Genet* 2013; 21(1): 14–20.
[20] UN Educational, Scientific and Cultural Organisation (UNESCO). 11 Nov 1997. Universal Declaration on Human Genome and Human Rights. Available at: http://portal.unesco.org/en/ev.php-URL_ID=13177&URL_DO=DO_TOPIC&URL_SECTION=201.html [Accessed 8 August 2016].
[21] NPRM for Revisions to the Common Rule, 45 C.F.R. 46 2015.
[22] H.F. Lynch, B.E. Bierer & I.G. Cohen. Confronting Biospecimen Exceptionalism in Proposed Revisions to the Common Rule. *Hastings Cent Rep* 2016; 46(1): 4–5.
[23] K.L. Hudson & F.S. Collins. Bringing the Common Rule into the 21st Century. *N Engl J Med* 2015; 373(24): 2293–2296.

THE GENOME AS COMMON

Having established the strongly held view that the genome is personal in nature, and the potential benefits to individuals and privacy protections that follow, we must turn our consideration to the common nature of the genome and the policies that can be derived from the shared aspect of DNA. In what sense is DNA 'shared' or 'common'? The most powerful sense in which our genome is 'common' lies in the fact that all people belong to the human gene pool. The human gene pool contains the set of genes of the whole human species. It is a repository for our genetic history, as well as the source of future generations.[24] Furthermore, as members of the human species, we all have a stake in what becomes of the human gene pool.

The stake that each person has in the human gene pool explains, at least in part, the widespread concern with human germline modification. Recent debates about preimplantation genetic diagnosis (PGD), which involves embryo selection, and mitochondrial donation, which involves embryo manipulation, highlight many people's heightened sense of caution when it comes to molding the human germline in any way. A 2016 Institute of Medicine (IOM) committee report on the ethical and social policy issues related to mitochondrial replacement techniques (MRT) considered the view that the human gene pool is a resource shared among the world's people, and therefore should not be modified without the consent of all humans.[25] The IOM Committee concluded, however, that given the small number of people who would utilize MRT, genetic changes would not take place at a scale capable of impacting the evolutionary processes of the gene pool. It would not be necessary, therefore, to consult each person before modifying the genome on such a small scale.

The IOM committee assumed that the only concern a person may have about the gene pool is consequentialist – that harmful evolutionary effects may occur downstream. It is possible, however, for someone to have an interest in the composition of the gene pool *per se*. Concern with the gene pool's makeup itself, however, has historically been associated with the eugenics movement. Francis Galton wrote in 1904 of improving the 'stock' of the human population.[26] The goal of ameliorating the quality of the common gene pool was used to justify forced sterilizations of those considered to have undesirable genes,[27] marriage restrictions based on race and class biases, and other degrading policies.[28] Eugenics represents the absolute repression of individual rights for the sake of achieving a so-called common ideal, and the reprehensible intellectual and political history of eugenics serves as a warning against taking the shared nature of the human genome to an inexcusable extreme.

Not all concerns with the makeup of the gene pool, however, result in eugenics. Some believe that there is both intrinsic and instrumental value in human genetic diversity. Instrumentally, the diversity of the human genome allows our species to be resilient, to adapt in the face of evolutionary pressures. Intrinsically, the diversity of the human gene pool is one of its most qualitatively rich features.[29] Furthermore, some argue from the disability rights perspective that human genetic diversity has important epistemic and moral value[30] and even that disability is an inherent part of the human condition.[31] Tolerance for and acceptance of human genetic variation is therefore an important human value to uphold, a value that may be undermined by efforts to select against certain heritable conditions using prenatal and preimplantation genetic testing.[32] Disability arguments are also posed in both instrumental (avoiding harm) and intrinsic (respecting diversity for its own sake) terms, but the central claim is that our attitudes towards the makeup of the human gene pool are morally significant and relevant to all.

The gene pool concept has been adapted by a variety of ideologues to achieve better or worse moral aims – indeed, even aims that are diametrically opposed (e.g. to subvert or buttress disability rights). But despite its pliant nature, the underlying claim of the gene pool concept remains constant and rings true; the human gene pool is common to all people, and membership of the human gene pool justifies a universal interest in what becomes of the gene pool.[33] Different interpretations and

[24] M. Queloz, *op. cit*. note 3.

[25] Institute of Medicine. 2016. *Mitochondrial Replacement Techniques: Ethical, Social, and Policy Considerations*. Available at: http://www.nap.edu/catalog/21871/mitochondrial-replacement-techniques-ethical-social-and-policy-considerations [Accessed 14 Apr 2016].

[26] F. Galton. Eugenics: Its Definition, Scope, and Aims. *Am J Sociol* 1904; 10(1): 1–25.

[27] In addition to making the case for cleansing the common gene pool for the sake of genetic purity, eugenicists also argued in consequentialist terms that their methods of removing undesirable genes would reduce the prevalence of crime and degenerate behavior. There were thus both intrinsic and instrumental reasons for pursuing eugenics from the eugenicist's point of view.

[28] A. Buchanan. Institutions, Beliefs and Ethics: Eugenics as a Case Study. *J Polit Philos* 2007; 15(1): 22–45.

[29] M. Queloz, *op. cit*. note 3.

[30] R. Garland-Thomson The case for conserving disability. *J Bioeth Inq* 2012; 9(3): 339–355.

[31] S. Woodcock. Disability, Diversity, and the Elimination of Human Kinds. *Soc Theor & Pract* 2009; 35(2): 251–278.

[32] E. Parens & A. Asch. Disability rights critique of prenatal genetic testing: reflections and recommendations. *Ment Retard Dev Disabil Res Rev* 2003; 9(1): 40–47; A. Asch & D. Wasserman. 2014. Reproductive Testing for Disability. In *The Routledge Companion to Bioethics,* J.D. Arras, E. Fenton & R. Kukla, eds. New York, Routledge: 417–432.

[33] Ossorio, *op. cit*. note 3.

implications of 'what becomes of the gene pool' will be considered in the next section.

Another sense in which DNA is common is literal; we share 99.9% of our DNA with other human beings.[34] (This means our genomes differ by approximately six million out of six billion nucleotides.) There does not seem to be much normative significance in this fact, however, since we also share 98.8% of DNA with chimpanzees and 90-95% of our DNA with mice. The number 99.9% does not, in itself, appear to be morally relevant. While we do typically assign greater moral status, and legal protections, to non-human animals that are more closely related to humans genetically, it is not because of their genetic similarity *per se*, but because of their enhanced capacities – for instance, for pleasure, pain or cognition.[35] Justifying a certain kind of treatment of non-human animals on the basis of the amount of DNA they have in common with humans is like justifying a certain level of obligation to close relatives on the basis of their genetic similarity; while we may owe more to those with closer genetic ties, our obligations do not emanate from our genes.

While the gene pool concept provides a more substantively meaningful sense in which the genome is common to all people, it is actually the literal commonality (99.9% of shared DNA), combined with the diversity created by genetic mutation, that makes large-scale genomic databases useful tools capable of bringing health benefits to all.[36] Large samples of genetic data can help scientists discover gene-disease associations by identifying variants that contribute to disease. Furthermore, large, representative studies can help us define and distinguish between genetic and environmental factors that contribute to health and disease, resulting in treatments for common illnesses.[37] With greater understanding of the causes of disease, health promotion and disease prevention programs can be specifically targeted towards susceptible individuals and populations based on their 'genomic profile and risk stratification'.[38] The common nature of the genome is thus not only an underappreciated theoretical framework, it is also the grounding for important advances and interventions that can improve health on a population scale.

THE GENOME AS PUBLIC

Thus far, the common nature of the human genome has been used to explain why, as members of the human gene pool, people share an interest in what becomes of the genome, and how the common aspect of the genome has possible implications for policies on germline modification and large-scale health innovations. However, not everything that people have in common necessarily falls into a domain that public representatives have the authority to govern. For instance, language is an important mediator of common human interaction, but except for extreme circumstances, the makeup and use of language are considered outside governmental control. Other examples of common phenomena generally believed to be beyond the reach of governmental authority include scientific knowledge and the arts. Thus in order for public health officials to have the license to develop policies on genetics issues, the genome must not only be *common*, but *public*. The next sections will explore two different frameworks for transitioning from *common* to *public* in the health and genomic context, along with the policy implications that result.[39]

Common heritage (CH) framework

One conceptual foundation that has been offered to undergird the public nature of the human genome is the notion of 'common heritage.' The Universal Declaration on the Human Genome and Human Rights opens with the statement that 'The Human Genome underlies the fundamental unity of all members of the human family, as well as the recognition of their inherent dignity and diversity. In a symbolic sense, it is the heritage of humanity.'[40] As legal scholar Pilar Ossorio writes, 'it seems eminently sensible to think of the human genome as part of our common heritage,' since, after all, 'Human genes are literally passed between generations; they unite each person with her and his forebears, descendants, and siblings,

[34] National Human Genome Research Institute. 2011. Whole Genome Association Studies. Available at: https://www.genome.gov/17516714 [Accessed 14 Apr 2016].

[35] B. Bastian, S. Loughnan, N. Haslam & H.R. Radke. Don't mind meat? The denial of mind to animals used for human consumption. *Pers Soc Psychol Bull* 2012; 38(2): 247–256; D. Degrazia. Human-animal chimeras: human dignity, moral status, and species prejudice. *Metaphilosophy* 2007; 38(2–3): 309–329.

[36] UN Educational, Scientific and Cultural Organisation (UNESCO). 11 Nov 1997. Universal Declaration on Human Genome and Human Rights. Available at: http://portal.unesco.org/en/ev.php-URL_ID=13177&URL_DO=DO_TOPIC&URL_SECTION=201.html [Accessed 8 August 2016].

[37] F.S. Collins. The case for a US prospective cohort study of genes and environment. *Nature* 2004; 429(6990): 475–477.

[38] A.M. Brand & N.M. Probst-Hensch. Biobanking for epidemiological research and public health. *Pathobiol* 2007; 74(4): 227–238.

[39] A more general discussion of how 'public' is defined and how it relates to governmental authority is beyond the scope of this article. See, for instance, Hannah Arendt's description of the public realm in Section II Chapter 7 of *The Human Condition*. Arendt equates the public realm to a common world that 'gathers us together and yet prevents our falling over each other' (p. 52). See also Jeff Weintraub's analysis of the public and private distinction in Chapter 1 of *Public and Private in Though and Practice: Perspectives on a Grand Dichotomy* (eds. Jeff Weintraub and Krishan Kumar), in which he contrasts the private, which 'pertains only to an individual' with the public, which 'is collective, or affects the interests of a collectivity of individuals' (p. 5).

[40] UNESCO, *op. cit*. note 20.

and they represent a connection among all human beings as members of the 'human family'.'[41] While there is no *single* genome that is common to all people, the collection of distinct, individual genomes, or gene pool, is 'a record of biological history and a source of future innovation' shared by all people.[42]

The CH concept, through comparison to other forms of common heritage (e.g. natural[43] or cultural heritage), transforms the genome from *common* to *public* by establishing that not only each person, but the collective – humankind at large – has an interest in what becomes of the genome. The Convention for the Protection of the World Cultural and Natural Heritage states that 'deterioration or disappearance of any item of the cultural or natural heritage constitutes a harmful impoverishment of the heritage of all the nations of the world' and that 'parts of cultural or natural heritage are of outstanding interest and therefore need to be preserved as part of the world heritage of mankind as a whole.'[44] Degrading an object of common heritage brings harm to humankind as a whole and the public has an interest in preventing such collective harms through the use of legal protections and prohibitions.[45]

The policies that flow from classifying the human genome as CH have two primary aims: preservation and access.[46] 'Preserving' the human genome can have several meanings. For instance, some believers in eugenics thought they were preserving the human gene pool by limiting its membership to those who represent the best of humanity. Preserving only a select portion of human genes, however, seems antithetical to the purpose of the CH concept. Others believe that any form of human germline manipulation constitutes an inappropriate, hubristic effort at 'playing God'.[47] It is not our place, they contend, to exercise control over our genetic makeup; the human gene pool should be preserved as it is, the way God made it.[48] In non-religious terms, one could argue that being products of natural evolutionary processes is essential to our humanity in some way, and tampering with the course of evolution detracts from our essential nature. Furthermore, there may be detrimental long-term consequences of using techniques that modify the germline that we cannot fully predict. The CH principle requires that we treat any manipulation of the human genome as an issue that affects all of humanity[49] and so it seems reasonable to adopt a cautious attitude towards technologies that introduce permanent changes in the gene pool.

In practice, then, preserving the human genome is not about purposefully shaping the makeup of the gene pool but rather about taking a cautious approach towards deliberate modifications to the germline and carefully regulating technologies that can be used to influence the makeup of the gene pool. These techniques include PGD, mitochondrial donation, and CRISPR/Cas9 gene editing. It is not necessary (or advisable) to impose an outright ban on these techniques in the name of genomic preservation, particularly given the small scale on which any of these techniques will be used. However, our society, through public deliberations, expert committees, or representative government action, should actively examine the circumstances and manner in which these technologies should be used. For instance, what constitutes a sufficiently severe genetic condition to warrant the use of PGD or MRT? Should MRT only be used on male embryos, so that the modifications to the mitochondria, which can only be passed down maternally, will not affect future generations? These and other questions should be addressed in a publically accessible, deliberative manner in order to achieve the goal of preserving the human genome without imposing norms about which genes belong in the human gene pool and which do not.

In addition to preservation, the CH conception of the human genome can also justify policies regarding access

[41] Ossorio, *op. cit*. note 3.
[42] Queloz, *op. cit*. note 3.
[43] The CH concept was first invoked in international law in the context of the seabed and ocean floor. Four elements of CH were enunciated: non-appropriation, international management, sharing of benefits, and reservation for peaceful purposes. These elements were designed to ensure that the seabed would be preserved for future generations and would remain accessible to all. (United Nations, Convention on the Law of the Sea, International Legal Materials, Treaties and Agreements. 10 Dec 1982. Available at: http://www.un.org/depts/los/convention_agreements/texts/unclos/unclos_e.pdf [Accessed 8 August 2016].)
[44] UN Educational, Scientific and Cultural Organisation (UNESCO). 16 Nov 1972. Convention Concerning the Protection of the World Cultural and Natural Heritage. Available at: http://whc.unesco.org/en/conventiontext/ [Accessed 14 April 2016].
[45] L. Gostin. 2000. *Public Health Law: Power, Duty, Restraint*. London, England: University of California Press: 88.
[46] In her paper 'The Human Genome as Common Heritage: Common Sense or Legal Nonsense?', Ossorio examines two conceptual historical and legal understandings of the the common heritage principle, the Common Heritage Property Doctrine (CHPD) and the Common Heritage Duties Doctrine (CHDD). The CHPD treats the genome as public property while the CHDD establishes common duties to preserve the human genome. This article will not engage directly with Ossorio's two conceptions of common heritage, since both can be used to justify policies related to preservation and access. Historically, the CHPD emphasizes fair distribution of genome-related resources and the CHDD highlights equal rights to access the genome, though it appears as though either the CHPD or CHDD could theoretically be used to justify both types of policies.
[47] Institute of Medicine, *op. cit*. note 25, p. 90.
[48] One could argue that modern medicine already alters the human gene pool by allowing people who would have died before having children to live longer and, in some cases, to pass deleterious genes on to the next generation. However, this is an example of the doctrine of double effect. The primary goal of the intervention is to save or prolong the life of the sick patient, thus although passing on deleterious genes to the next generation is a foreseeable consequence, the intervention is ethically permissible nonetheless.
[49] Ossorio, *op. cit*. note 3.

to genes and genomic information. If the genome is our common heritage, there should be fair access to genomic data.[50] The genome should not be monopolized by a few – whether by commercial entities, research programs or privileged nations. For example, many have posited that there should be international sharing of genomic data collected in regional or national biobanks.[51] Global sharing of genomic data can also be justified using utilitarian reasoning, since larger pools of genomic information will result in greater research opportunities and public benefits. However, beyond consequentialist logic, the notion that genomic information ought to be shared because the human genome is the common heritage of humankind provides a powerful reason for international data-sharing.

The implications of the CH concept for gene patenting are less clear. It may seem intuitive that if the human genome is common heritage, no individual, institution or company should be able to claim ownership over human genetic material. Moreover, in some countries, patents can be denied on moral grounds, for example if granting a patent would be 'contrary to public policy or morality.'[52] Though gene patenting has been widely allowed, one could argue that granting ownership of common human heritage would stand in opposition to public policy or morality. However, certain derivatives of DNA, which are not identical to naturally-occurring DNA and are fabricated by human beings, should not be considered part of the human gene pool and may therefore be patent-eligible under the CH concept.[53] Furthermore, through the patent law system, national or global communities can legitimately bestow ownership of genetic material.[54] It may actually be beneficial overall to allow temporary ownership of genetic material in order to encourage research and foster innovation (though introducing financial incentives for medical research can also result in conflicts of interest and research bias).[55] Nonetheless, the CH concept can at least be employed to support a default position of non-ownership of the human genome, with exceptions made for clear instances of human ingenuity or opportunities for substantial public benefit.

The CH approach and its policy implications highlight the inherent qualities of the human genome. The gene pool ought to be protected and preserved because humankind has an interest in its maintenance, due to its own intrinsic value. Global genomic data, in theory,[56] ought to be accessible to everyone because all people have a claim to the collective heritage of humanity. The next section will address some of the instrumental qualities of the human genome, building on a distinct notion of the public nature of DNA.

Common resource (CR) framework

According to the CR concept, the human genome, as a common resource, is a global public good. In classical economic terms, it is non-excludable and non-rivalrous. It is comparable to an environmental resource, from which all people can benefit without detracting from the benefit of others. Under the CR framework, the human genome is considered public not because of its inherent collective value, but because all people have an interest in the benefits derived from the resource – benefits brought about by proper resource management. The United Nations'Convention on the Law of the Sea, which was founded on the CH principle but also addresses the use and management of marine resources, states that 'exploration and exploitation' of the ocean and its resources 'shall be carried out for the benefit of mankind as a whole.'[57] Thus, like an environmental resource, the genome ought to be managed by public entities in order to protect the shared resource and maximize public benefit.[58] The genome is a shared asset that can be used to bring about scientific and medical developments that benefit all of humankind.[59] It is the genome's instrumental value to humanity that renders it public and justifies public policies regarding its use.

One policy that stems from the CR concept also relates to access to genomic data.[60] The International Ethics Committee of the Human Genome Organization (HUGO) has declared that human genomic databases

[50] B.M. Knoppers et al. A human rights approach to an international code of conduct for genomic and clinical data sharing. *Hum Genet* 2014; 133(7): 895–903.

[51] B.M. Knoppers et al. Towards a data sharing Code of Conduct for international genomic research. *Genome Med* 2011; 3(7):46; J.R. Harris et al. Toward a roadmap in global biobanking for health. *Eur J Hum Genet* 2012; 20(11): 1105–1111.

[52] UK Parliament. 1 Oct 2014. *The Patent Act of 1977 (as amended)*. Available at: https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/354942/patentsact1977011014.pdf [Accessed 13 Aug 2016].

[53] This position is consistent with the US Supreme Court's decision in the 2013 *Myriad Genetics* court case. The majority held that naturally occurring DNA is not patent-eligible, but complementary DNA (cDNA) is created by human beings and therefore can be patented.

[54] M. Queloz, *op. cit*. note 3.

[55] B. Capps. Can a good tree bring forth evil fruit? The funding of medical research by industry. *Br Med Bull* 2016; 118(1): 5–15.

[56] In practice, data security and privacy considerations will likely require limiting access to global genomic data, perhaps to those with the training and clearance to handle such sensitive information. However, anyone would theoretically be capable of gaining the skills and permission to access global genomic data, which follows from the CH concept.

[57] United Nations, Convention on the Law of the Sea, International Legal Materials, Treaties and Agreements. 10 Dec 1982. Available at: http://www.un.org/depts/los/convention_agreements/texts/unclos/unclos_e.pdf [Accessed 14 April 2016].

[58] P.N. Ossorio, *op. cit*. note 3.

[59] M. Queloz, *op. cit*. note 3

[60] B.M. Knoppers et al. Framing genomics, public health research and policy: points to consider. *Public Health Genomics* 2010; 13(4): 224–234.

should be considered global public goods because they are goods 'whose scope extends worldwide, are enjoyable by all with no groups excluded, and when consumed by one individual, are not depleted for others.'[61] Relatedly, if the human genome is a common resource, the benefits derived from genomics research should be shared equally around the world.[62] The Universal Declaration on the Human Genome and Human Rights emphasizes the need for benefit-sharing between developed and developing countries, stating that 'Benefits from advances in biology, genetics and medicine, concerning the human genome, shall be made available to all…'[63] Like the CH concept, the CR concept can be used to justify equitable access to the genome and the benefits resulting from genomics research.

Beyond equitable access, the view of the human genome as a common resource also justifies *using* the genome for the public interest. As with environmental resources, sound stewardship of the public resource often involves utilizing the good to maximize public benefit.[64] In the case of the human genome, as discussed earlier, large databases of genetic information can be employed to ascertain the genetic and environmental causes of disease and used to target public health interventions towards at-risk populations. Such databases carry tremendous potential for public benefit, including possible breakthroughs in medicine and public health.[65] Given the opportunities for public benefit, proper management of the human genome should include harnessing the power of the genome to maximize public gains made through genomics research (with some constraints, discussed later).

Recognizing the human genome as a common resource not only supports a mandate for those responsible for managing the public good, it also asserts a corresponding public interest in the common resource. Notably, this kind of public interest differs from the public interest stemming from the CH concept. The CH public interest is an interest in the sense that people *care about* and *identify with* what becomes of the genome, while the CR public interest is *material*. In other words, humankind has a claim to the public resource and is entitled to benefit from the products of genomic research.

In certain cases, this material public interest in the human genome will stand in tension with the private interest in the genome. For example, amassing large-scale biobanks is a prerequisite to maximizing the public health benefits of population-level genomics research, since the usefulness of genomic databases is directly linked to their comprehensiveness;[66] the more comprehensive the database, the more representative the research results will be for the population as a whole, particularly for racial and ethnic minorities.[67] To date, nearly all participation in national biobanking efforts has been voluntary, with individuals providing informed (and usually broad) consent to contribute their DNA given certain parameters on privacy, identifiability and the research that will be conducted.[68] However, based on the CR framework, an argument can be made for stronger public action aimed at recruiting participants to genomic databases, with the ultimate goal of harnessing population-wide genomic data to improve public health.

One such action would be to create a national biobank for genomic information without obtaining the explicit informed consent of participants. This was Iceland's approach; Iceland passed a law establishing a national biobank, which included genetic and other health-related information,[69] by incorporating previously-collected samples into the biobank with the *presumed* consent of individuals. There was an option to opt out, but it was limited to the first six months of incorporation into the database.[70] Iceland was able to pass this law because there was vigorous public debate beforehand and support of 75% of the population at the time that the law was passed.[71] Those who supported the Icelandic biobank contended that using the presumed consent standard made data collection significantly easier and improved

[61] Human Genome Organisation (HUGO), Ethics Committee: Statement on human genomic databases, December 2002. *J Int Bioethique* 2003; 14: 207–210.

[62] B.M. Knoppers et al., *op. cit*. note 50; United Nations, Convention on Biological Diversity. *The Nagoya Protocol on Access to Genetic Resources and the Fair and Equitable Sharing of Benefits Arising from their Utilization to the Convention on Biological Diversity*. 29 Oct 2010. Available at: https://www.cbd.int/abs/text/ [Accessed 12 Aug 2016].

[63] UNESCO, *op. cit*. note 20.

[64] W. Burke et al. Extending the reach of public health genomics: what should be the agenda for public health in an era of genome-based and 'personalized' medicine? *Genet Med* 2010; 12(12): 785–791; F.S. Collins et al., US NHGRI. A vision for the future of genomics research. *Nature* 2003; 422(6934): 835–847.

[65] J.R. Harris et al., *op. cit*. note 51; H. Swede, C.L. Stone & A.R. Norwood. National population-based biobanks for genetic research. *Genet Med* 2007; 9(3): 141–149.

[66] B.M. Knoppers et al., *op. cit*. note 50.

[67] See, for example, C.D. Bustamante, E.G. Burchard & F.M. De la Vega. Genomics for the world. *Nature* 2011; 475(7355): 163–165; for a discussion of the fact that most genome-wide association studies are highly skewed towards people of European descent and the consequences for population genomics.

[68] M.A. Rothstein, B.M Knoppers & H.L. Harrell. Comparative Approaches to Biobanks and Privacy. *J Law Med Ethics* 2016; 44(1):161–172; H. Swede et al., *op. cit*. note 65.

[69] M.G. McInnis. The assent of a nation: genethics and Iceland. *Clin Genet* 1999; 55(4): 234–239.

[70] Althingi. Act on a Health Sector Database No. 139/1998. Available at: https://eng.velferdarraduneyti.is/media/acrobat-enskar_sidur/Act-on-a-Health-Sector-Database-as-amended.pdf [accessed 16 Aug 2016].

[71] H. Swede et al., *op. cit*. note 65.

the quality of the database, since it would be more representative of the national population.[72] However, due to a combination of financial instability, public disapproval in Iceland regarding the involvement of a commercial company,[73] and international skepticism regarding the presumed consent model, the database was never fully realized.[74] Thus despite the strong initial support for a national database in Iceland, pragmatic and ethical concerns prevented the database from achieving its public health potential.

In the United States, there is substantial debate surrounding the question of whether existing samples should be made available for public health research without the explicit consent of participants.[75] Some empirical data shows a preference for opt-in versus opt-out consent models for using biological samples in secondary research,[76] particularly with regards to parental consent to research on residual newborn screening bloodspots.[77] However other surveys show that many people would be willing to participate in research under an opt-out system[78] and approved of existing opt-out biobanking efforts, such as Vanderbilt University's BioVU biorepository (which has since switched to an explicit consent process).[79] Nevertheless, the NPRM for revision of the Common Rule proposes that informed consent be required for secondary research with a biospecimen, even if the specimen is de-identified.[80] After all, biospecimens can be used to generate genetic information, which is unique to individuals and cannot truly be de-identified.[81] The NPRM leaves room for a limited set of exemptions, including public health surveillance, research conducted by a government agency using government-collected data, and secondary research use of identifiable private information originally collected as part of a non-research activity, where notice of such possible use was given.[82] Precisely what activities fall under these exemptions has yet to be determined, but exemptions are meant to be kept to a minimum. In general, the NPRM represents a move towards obtaining informed, if broad, consent for secondary research use, and away from opt-out consent models.

The view of the human genome as a common resource pushes back against efforts to tighten regulations for public health research on stored genomic data. Though respect for the autonomy of participants is important, it is not the only factor that must be considered in deciding whether conducting secondary research is worth the potential risks to individuals. There is also a public interest in the human genome, public entitlement to the benefits of genomics research, and public health mandate to maximize health gains through carrying out research on the genome.[83] The revision to the Common Rule presents an opportunity for American genomics policy to reflect a wider set of conceptions of the human genome's value – one that includes the genome's intrinsic and instrumental value to the population as a whole. In its current form, the NPRM disproportionately reflects the personal nature of DNA and limits our ability to maximize the public health benefits of genomics research.

It is important to remember, however, that the success of public health measures often depends on maintaining the public trust. If large portions of the population oppose conducting genomics research on stored samples, it would be counterproductive to forge ahead, potentially causing long-term damage to the public's trust in public health agencies. Iceland's example, while ultimately unsuccessful and on a relatively small scale, demonstrates that it may be possible to establish public support for a nationwide opt-out genomics database through public engagement in decision-making and efforts to educate the public on the risks and benefits of the genomics resource. Iceland's example also demonstrates the importance of maintaining the public trust throughout the implementation process, not only when garnering initial

[72] M.G. McInnis, *op. cit*. note 69.

[73] S.B. Haga & L.M. Beskow. Ethical, legal, and social implications of biobanks for genetics research. *Adv Genet* 2008; 60: 505–544.

[74] D. Winickoff. 2015. A Bold Experiment: Iceland's Genomic Venture. In *Ethics, Law and Governance of Biobanking*. D. Mascalzoni, ed. Springer: 187–209.

[75] M. Bayefsky, K. Saylor, & B. Berkman. Parental Consent for the Use of Residual Newborn Screening Bloodspots: Respecting Individual Liberty vs. Ensuring Public Health. *JAMA* 2015; 314(1): 21–22; N.A. Garrison et al. A systematic literature review of individuals'perspectives on broad consent and data sharing in the United States. *Genet Med* 2016; 18(7): 663–671.

[76] C.M. Simon et al. Active choice but not too active: public perspectives on biobank consent models. *Genet Med* 2011; 13(9): 821–831.

[77] B.A. Tarini et al. Not without my permission: parents'willingness to permit use of newborn screening samples for research. *Public Health Genomics* 2010; 13(3): 125–130; J.R. Botkin et al. Public attitudes regarding the use of residual newborn screening specimens for research. *Pediatrics* 2012; 129(2): 231–238.

[78] D. Kaufman et al. Preferences for opt-in and opt-out enrollment and consent models in biobank research: a national survey of Veterans Administration patients. *Genet Med* 2012; 14(9): 787–794.

[79] K.B. Brothers, D.R. Morrison & E.W. Clayton. Two large-scale surveys on community attitudes toward an opt-out biobank. *Am J Med Genet* 2011; 155A(12): 2982–2990.

[80] NPRM, *op. cit*. note 21.

[81] K.L. Hudson & F.S. Collins, *op. cit*. note 23.

[82] NPRM, *op. cit*. note 21.

[83] Knoppers et al. argue that personal health information, including genetic data, held in existing databases ought to be available for secondary public health research when 1) accessing such data is deemed necessary for research, 2) re-contacting individuals for new consent is impractical or unfeasible, 3) there are appropriate mechanisms in place for data security, and 4) the risk of identifiability is outweighed by the benefits of research (B.M. Knoppers et al., *op. cit*. note 60.). While determining exactly how to weigh the privacy risks against the benefits of research remains an open question, these authors support the claim that the public interest in genomics research is legitimate and carries moral weight when developing genomics policy on national biobanking.

support. As a Nuffield Council report on public health data initiatives states, efforts should be made to learn about the public's expectations regarding the use of their genomic data, and decision makers must continuously 'engage with those expectations'.[84]

In particular, public health agencies would need to elucidate the privacy protections that will be put in place and provide a realistic assessment of the likelihood that samples will be re-identified. Furthermore, significant effort must be made to ensure that the genomic data are as secure as possible. Public health officials should also formulate and publicize the kinds of research that will be carried out, in order to persuade people that participation is in the public interest. Equally important, the public must be aware of applications that would undermine public trust that will *not* be pursued, such as the use of DNA stored in the national biobank for criminal investigations. Finally, in order to ensure that acting in the public interest does not preclude respect for individual autonomy, there should be an option to opt out of the database for those who perceive the individual risks to be too great or oppose another aspect of the biobanking project. Public health agencies should also undertake to understand the concerns of those who decide to opt out, and seek to address those concerns to the extent possible.

The notion of the human genome as a common resource provides a conceptual framework for maximizing the instrumental value of the human genome. All of humankind has an interest in the genome and proper management of the common resource involves utilizing the genome to benefit the public – even if this requires adopting a stronger stance towards recruitment to population-level genomics databases. Furthermore, the CR concept supports equitable sharing of the benefits of genomics research among all people.

CONCLUSION

Ultimately, harnessing the power of the human genome to maximize public benefit requires balancing respect for individuals with the promotion of common aims. This article has focused on the latter because the promotion of public health goals through DNA biobanking has received insufficient consideration in the literature surrounding the ethics of genomics research. However, any large-scale DNA biobanking effort must include privacy protections, data security, and recourse for individuals who strongly desire to opt out – in order both to respect individual autonomy and to maintain the public trust. Similarly, policies designed to preserve and protect the human genome should balance public and private interests. For example, policies on mitochondrial donation should weigh the needs of individuals and families with heritable mitochondrial disease against the public interest in preservation of the germline. Neither private nor public interests take clear precedence in developing policies on genetics issues, but they must be weighed against each other and accommodate each other when possible.

The CH and CR frameworks establish a public interest in the human genome based on the intrinsic and instrumental properties of DNA. Since acting in the public interest on health issues is typically seen as the role of public health agencies, the CH and CR concepts most easily lend themselves to a top-down approach to genetics policy development. However, the common nature of the human genome should also provide a reason for individuals to voluntarily contribute to efforts to maximize the public benefits of genomics research, drawing upon the principle of solidarity.[85] A Nuffield Council report on solidarity in bioethics explains that a solidarity-based view of biobanking assumes 'that when individuals decide to participate in biobank-based research, they are willing to accept the possibility that a certain level of costs may need to be carried by them for the sake of communal benefit.'[86] A further argument could be developed that individuals actually have a *duty* to work towards preserving and utilizing the human genome. These top-down and bottom-up approaches are complementary, and pursuing both approaches simultaneously will ensure that public health agencies are not acting in the public interest, paradoxically, against the public's will. To avoid this paternalistic outcome, educational campaigns and public participation should be part of any process to develop policy on genetics issues.

This article aimed to provide a conceptual underpinning to the public interest in the human genome and to use the common nature of DNA to justify policies on preserving and harnessing the genome. Additional work is needed to understand exactly how to weigh the public benefits against the privacy risks to individuals for a particular genomics policy. Furthermore, this article took for granted the premise that striving to preserve the human genome is a laudable goal given the inherent value of the genome. It is not obvious, however, that preservation is the only or correct response to valuing the human genome for its own sake, or precisely what preservation entails. Finally, this paper does not address the level at which genetics policy ought to be developed – at a regional, national or international level. The conception

[84] Nuffield Council on Bioethics. 2015. *Ethical governance of data initiatives.* Available at: http://nuffieldbioethics.org/report/collection-linking-use-data-biomedical-research-health-care/ethical-governance-of-data-initiatives/ [accessed 12 Aug 2016].

[85] R. Chadwick & K. Berg. Solidarity and equity: new ethical frameworks for genetic databases. *Nat Rev Genet* 2001; 2(4): 318–321.

[86] B. Prainsack & A. Buyx. 2011. *Solidarity: Reflections on an emerging concept in bioethics.* Wiltshire: ESP Colour Ltd.

of the genome as the common heritage of humanity, or a common resource shared by all humans, most directly supports biobanking at an international level.[87] However, international policy development on genetics issues will also need to take into account geopolitical considerations that are beyond the purview of this article.

Although the personal and private nature of the human genome is undeniable, there is also an unmistakable common and public aspect of our DNA that should be taken into account when creating policies on genetics. Just as individuals have an interest in genetic privacy, the public has a legitimate interest in the makeup and use of the human genome. Developing genetics policies will be a challenging venture in pursuing the public interest by maximizing public health benefits, respecting individual autonomy, and engaging the public in an open and deliberative process.

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Disclaimer

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[87] B.M. Knoppers. Biobanking: international norms. *J Law Med Ethics* 2005; 33(1): 7–14.