HHS Public Access

Author manuscript

Genet Med. Author manuscript; available in PMC 2015 December 04.

Published in final edited form as:

Genet Med. 2015 December ; 17(12): 949–957. doi:10.1038/gim.2015.8.

Stakeholder engagement in policy development: challenges and opportunities for human genomics

Amy A. Lemke, MS, PhD[1] and **Julie N. Harris-Wai, PhD, MPH**[2,3]

[1]Center for Genomics and Healthcare Equality, University of Washington, Seattle, Washington, USA

[2]Division of Research, Kaiser Permanente, Oakland, California, USA

[3]Institute for Health and Aging, University of California, San Francisco, San Francisco, California, USA

Abstract

Along with rapid advances in human genomics, policies governing genomic data and clinical technologies have proliferated. Stakeholder engagement is widely lauded as an important methodology for improving clinical, scientific, and public health policy decision making. The purpose of this paper is to examine how stakeholder engagement is used to develop policies in genomics research and public health areas, as well as to identify future priorities for conducting evidence-based stakeholder engagements. We focus on exemplars in biobanking and newborn screening to illustrate a variety of current stakeholder engagement in policy-making efforts. Each setting provides an important context for examining the methods of obtaining and integrating informed stakeholder voices into the policy-making process. While many organizations have an interest in engaging stakeholders with regard to genomic policy issues, there is broad divergence with respect to the stakeholders involved, the purpose of engagements, when stakeholders are engaged during policy development, methods of engagement, and the outcomes reported. Stakeholder engagement in genomics policy development is still at a nascent stage. Several challenges of using stakeholder engagement as a tool for genomics policy development remain, and little evidence regarding how to best incorporate stakeholder feedback into policy-making processes is currently available.

Keywords

genomics; key stakeholders; policy; policy making; stakeholder engagement

Along with rapid advances in human genomics, policies governing genomic data and clinical technologies have proliferated. We broadly refer to “genomics policy” as formal action plans or principles to guide decision making and best practice in research, public health, and clinical care. Genomics policies at federal, state, organization, and institutional

Correspondence: Amy A. Lemke (aalemke@wisc.edu).

DISCLOSURE

The authors declare no conflict of interest.

levels address a wide spectrum of policy issues, from how genomics research samples are stored and shared to whether and how to utilize new genomics technologies in clinical practice.

The Institute of Medicine and key constituents in genomics acknowledge the value of engaging key stakeholders in the development of clinical and research policies to create sound, transparent, and trusted health policy.[1–6] In addition, stakeholder involvement in patient-centered care, patient-centered outcomes research, and evidence-based health policy decision-making efforts is increasingly recognized as "contextual evidence" that informs clinical practice, research, and policy.[7–13] While there is wide agreement about the need for stakeholder engagement, few models or theoretical foundations guide how to design, conduct, evaluate, or use the outcomes of stakeholder engagement in the process of developing policy. Furthermore, detailed methods and outcomes of stakeholder engagement for policy development are rarely published. Therefore there is a dearth of empirical data on best practices for stakeholder engagement in the policy-development arena. With the complex and evolving landscape of genomics, some key questions should be examined. Who are key stakeholders in genomics and how are they involved in policy development? Why engage stakeholders in genomics policy issues? When are stakeholders engaged in the policy-development process? What are potential challenges in engaging stakeholders in genomics policy generation?

The purpose of this article is to examine how stakeholder engagement is used to develop policies and guidelines in specific genomics research and public health areas, as well as to identify future priorities for conducting evidence-based stakeholder engagements. We focus on exemplars in biobanking and newborn screening (NBS) to illustrate a variety of current stakeholder engagement policy-making efforts. Each setting provides an important context for examining the methods of obtaining and integrating informed stakeholder voices into the policy-making process.

WHO ARE KEY STAKEHOLDERS IN GENOMICS AND HOW ARE THEY INVOLVED IN POLICY DEVELOPMENT?

Broadly defined, a stakeholder is a person, group, or organization involved in or affected by a course of action. Key stakeholders in genomics include diverse groups of patients, research participants, the public, providers, researchers, advocacy groups, payers, policy makers, and others. Stakeholder engagement refers to the *process* by which an organization involves people who may be affected by the decisions it makes or who can influence the implementation of decisions. Stakeholders may support or oppose decisions and may be influential in the organization or within the community in which they operate. In this article we focus on stakeholder engagement methods and the integration of feedback throughout various genomics policy-development processes.

The type and extent of key stakeholder involvement in genomics policy development may depend on a number of factors, including the specific context of the policy development (e.g., large biobank, small state genomics program); the purpose of the engagement; available resources; and the power-sharing structure of the groups involved. Therefore the

engagement methods used will likely vary. Several different models describe a type of continuum, or different levels, of stakeholder involvement in decision making.[14,15] For example, the International Association of Public Participation's spectrum of participation defines five broad levels of increasing involvement in the engagement process: (i) inform (e.g., fact sheets, websites, open houses), (ii) consult (e.g., public comment, focus groups, surveys, public meetings), (iii) involve (e.g., workshops, deliberative polling), (iv) collaborate (e.g., citizen advisory committees, consensus building, participatory decision making), and (v) empower (e.g., citizen juries, delegated decisions).[15] We consider this spectrum of participation when assessing our case examples of stakeholder engagement in genomics policy decision making.

WHY ENGAGE STAKEHOLDERS IN GENOMICS POLICY ISSUES?

There are several reasons to consider involving key stakeholders in genomics policy development (Table 1). On a practical level, stakeholder engagement identifies areas of agreement as well as disagreement and provides an opportunity to understand more fully what might be driving key stakeholder differences. Stakeholder input may also help articulate the values of the broader community affected and align policy recommendations with these expectations. Genomics policy decisions tend to involve a large amount of complex, technical information that may be difficult for laypeople to comprehend.[16–18] If an educational component is provided, stakeholder engagement may also increase genetic literacy. In addition, by building mutual understanding, credibility, and trust, policies may be more likely to be implemented as intended by the key stakeholders.

Stakeholder input is also an important factor in increasing the quality and trustworthiness of health policy aimed at improving health-care quality and outcomes.[1] Mistrust over the use of genetic information could affect perceptions about, and participation in, research and clinical genomics services. In several recent high-profile cases, lawsuits were brought against genetics researchers[19] and public health programs,[20] and in two cases valuable bloodspots for use in research from state NBS programs were destroyed.[21–23] Current efforts, however, are increasingly recognizing the importance of including key stakeholder groups (family members, for example) in developing and implementing genomic policy, as evidenced by the National Institutes of Health involvement of the Lacks family in a genomic data access and governance policy.[24,25]

WHEN ARE STAKEHOLDERS ENGAGED IN THE POLICY-DEVELOPMENT PROCESS?

Although there is no perfect, one-size-fits-all model for developing policies or guidelines, defining stakeholder roles in any or all stages of genomics policy making is important to better evaluate and understand the policy-making process. A number of frameworks have been developed in various disciplines to assist policy makers in planning for policy development and analysis, and some include a specific component addressing key stakeholder consultation.[26,27] General stages of policy development typically include some elements of the following: agenda setting, analysis, policy formation, implementation, and policy review and evaluation. Figure 1 illustrates when stakeholders might be engaged at

various phases of the policy cycle. While these stages seem to be chronological, beginning with agenda setting and ending with implementation and evaluation, policy decision making in practice rarely follows a linear format. These stages may occur frequently throughout the lifetime of a single policy, and steps may be skipped—or reversed—along the decision-making process.

WHAT ARE POTENTIAL CHALLENGES IN ENGAGING STAKEHOLDERS IN GENOMICS POLICY GENERATION?

Stakeholder engagement in genomics policy generation presents several challenges. First, identifying key stakeholders that are representative of a larger group may be difficult. In genomics, for example, selecting members to represent “the public” or “patients” can be challenging because who will be affected by the technologies and who has a stake in the particular policy are often unclear. Furthermore, each individual stakeholder may have views that differ from others in their constituency. Therefore, defining and sharing the selection process for identifying key stakeholders is important. Second, stakeholder engagement processes require resources; the extent of those resources is dependent on the method of engagement chosen. For example, a deliberative engagement incurs more costs than a web conference, and some engagement strategies require specialized expertise, knowledge, and skills in those methods. Depending on the method used, there needs to be a certain amount of time (and funding) to plan, conduct, analyze, and disseminate the findings of stakeholder engagement. A major challenge in stakeholder engagement in genomics is determining how and when to incorporate key stakeholder input into policy decision making. Different interests and stakeholder cultures may be involved, which may cause difficulties in reaching agreement about a particular policy. Determining in advance how to deal with divergent views in policy making is an essential step in ensuring sound policies for any institution or program. Finally, there must be a well-articulated plan and process for informing stakeholders about how their input will be used or potentially not used. This helps ensure transparency and increase public trust and endorsement of (i) the process used to listen to and involve stakeholders and (ii) the resulting policy.

ENGAGEMENTS IN GENOMICS POLICY DEVELOPMENT

A wide range of policies and guidelines exist in human genomics to facilitate the translation of genomics information and technology that span federal, state, and local issues. Therefore the goals of stakeholder engagement vary to reflect specific circumstances, needs, and resources. Through two broad genomics examples (biobanking and NBS), we describe how key stakeholders are currently involved in different human genomics policy-making efforts. Although the extant literature on stakeholder engagements elicits views toward genomic issues in general, few studies focus on how engagement findings are incorporated into policy decisions. Therefore, we selected diverse cases that include not only stakeholder engagements but also examples that detail how the engagement findings inform specific genomics policies and that document outcomes of the engagement related to policy. Our descriptions are not intended to be a comprehensive or systematic review, but rather a set of select examples in genomics to demonstrate the diversity of organizations, types of key stakeholders engaged, engagement purposes, when the engagement occurred in the policy-

development cycle, methods of engagement, and outcomes related to policy (Tables 2 and 3). While some of these engagements may have used evaluations of stakeholder engagements in the policy-making process, specific evaluation methods and outcomes were not reported in the publications reviewed.

Biobanks

Biobanks, which store large amounts of participants' genomic DNA and other health data, are increasingly used by government, academic, and research organizations to fuel biomedical research, with the ultimate goal of improving public health. In part because of the diversity of biobanks and their missions, policies that govern the use of participant data lack uniformity.[28] Through engagement initiatives, stakeholders with a vested interest have been called on to provide input into institutional, state, and federal biobanking policies to inform a variety of practices in areas such as genomic data collection, storage, and sharing. In the biobank research community, however, there is variability in understanding what constitutes engagement, how to conduct the engagement process, and why engagement is conducted.[29] In addition, how the engagement findings are used in the development of actual policies is not always clear. The examples in Table 2 were selected to illustrate a range of current stakeholder engagement strategies used to inform specific biobank research policies.

The types of biobanks listed reflect mainly large-scale efforts to recruit from area populations; they are not specific to a disease category (Table 2). For example, the Mayo Clinic biorepository houses data from Minnesota residents, and Kaiser Permanente's Research Program on Genes, Environment, and Health has data from large numbers of Kaiser Permanente's Californian members. Not included in the examples are smaller-scale biobanks, those that collect and store disease-specific data, and those that are organized and governed by participants. All of these factors influence who is likely to be involved as key stakeholders and what drives the engagement purpose and approach related to a policy-making effort. Key stakeholders in the various examples listed include community members, participants, investigators, researchers, advisory board members, and clinicians. In the two examples of National Institutes of Health groups that are involved in broader genomics policy development, the reach to key stakeholders included researchers and patient communities involved with the specific disease (cancer), research participants, and the larger public.

The purposes of the different engagements reflect in part the particular stage in biobank development, as well as various interests such as assessing ongoing and new operating procedures, formulating best practices (National Cancer Institute), and informing national policy (genomic data sharing). Stakeholders were involved in different stages of policy development depending on these various goals. Kaiser Permanente (Research Program on Genes, Environment, and Health), the Mayo Clinic biobank, and the National Cancer Institute seemed to involve stakeholders at each stage of the policy cycle, possibly reflecting the ongoing nature of their operations and research. The international HapMap Project seemed to involve key stakeholders in the policy-formation and -implementation stages, reflecting their particular research goals at various time points.

A wide range of engagement methodologies were used by the various biobanks and organizations involved in genomics policy-making efforts. Again, this is not surprising because many of the engagement goals differed, and the methods matched the particular circumstances—and possibly the resources available. The biobanks used a variety of methods that involved different levels of stakeholder involvement, as described earlier in the example of the International Association of Public Participation decision-making participation continuum. Most of methods in Table 2 involved the levels of “informing” and “consulting with” their key stakeholders, which is reflected in the methods chosen, for example, website posts, public online comments, surveys, and public meetings. These approaches may have been chosen because they can be used fairly easily to get broad feedback regarding a new or existing policy (data sharing, for example). Moving up the spectrum to a higher level of stakeholder involvement, deliberative engagement was used as a method by both the Mayo Clinic and the BC BioLibrary to engage participants, and potential participants, in decision-making processes related to protocols, policies, and biobank governance. This method, however, requires more expertise and funding compared with the informing and consulting methods. An overall theme in the examples listed was the use of advisory groups or committees to guide and inform genomics policy decisions over time. How the input of these advisory groups was specifically used to inform or change policy was not reported.

All of the examples in Table 2 document some type of outcome(s) of their engagement related to policy. Many of the biobanks described specific policies that were implemented as a result of the engagement, such as the use of an opt-in mechanism for reconsent, the option for participants to withdraw from the biobank, and the implementation of a “permission to contact process” for potential donors to be contacted by the biobank to discuss additional study recruitment. An engagement outcome for the National Cancer Institute and the Mayo Clinic was the development of advisory groups (the Biorepository Coordinating Committee and the Community Advisory Board, respectively) to continue to provide practice and policy guidance. Lessons learned from the HapMap Project included understanding the importance of involving diverse groups to learn how they wished to have samples from their locality collected and described. An outcome from the HapMap stakeholder engagement was to label samples to reflect social identities and to use lay language in their Statement of Research Intent. Last, in the National Institutes of Health example, an outcome of the public draft comments and town hall meetings was the culmination of a final Genomic Data Sharing Policy in 2013, with the public comments posted online.

Newborn screening

Each state in the United States screens every newborn infant born in a hospital (many states cover births outside of hospitals) for a panel of different rare genetic or metabolic disorders.[30] Over the past 40 years, there has been a significant reduction in morbidity and mortality as a result of NBS programs.[31] Because these programs are deemed necessary to the health of developing infants, most states do not require or obtain explicit consent of the parents or guardians to participate in the program. NBS has come under examination in the past several years as the public has become more aware of NBS and the various uses of the leftover screening bloodspots in research.[32,33] Therefore, several state, national, and

international NBS programs have attempted to engage and/or educate community members about NBS programs in an effort to set policy.

Engagements have been conducted to inform various policies such as research use of leftover bloodspots as well as the potential for expanding the NBS panel because of increasing evidence about certain disorders and/or technological advances (e.g., whole-genome and whole-exome sequencing). A variety of these engagements are listed in Table 3 to illustrate the types of stakeholders that are involved, the unique purposes of the engagement, as well as the corresponding stage of the policy-development process when engagement occurred, methods used, and outcomes of the engagement related to policy. We highlight stakeholder engagements for national NBS policies as well as individual state policies to show the diversity of policy decisions within a single context (e.g., NBS).

As shown in Table 3, key stakeholders in the NBS engagements listed were usually reported as patients or parents of children with rare disorders, disease advocates or advocacy groups, and medical and scientific experts. The one exception was the Michigan BioTrust for Health, where stakeholders were identified as individuals from the lay public. The purpose of the Michigan engagement was to gather feedback regarding the ongoing biorepository, whereas the purposes of the other engagements were mainly to obtain guidance, or a framework, for determining what conditions/tests to add to or discontinue from the NBS panel. Noting that the Michigan BioTrust program could have been interpreted as a biobanking example or a NBS example. Because the BioTrust program addresses the use of leftover bloodspots from a NBS program and most NBS programs are facing significant policy challenges with respect to the use of leftover bloodspots, we chose to include it as an example in the NBS context. The engagement for the Department of Health and Human Services was broader in that it focused on obtaining guidance on all aspects of the NBS program (e.g., parent education, use of leftover bloodspots, and screening panel criteria).

Determining the stage at which any engagement operates is difficult because there is often limited documentation on how engagement efforts are integrated into decision making. In the examples in the tables, Michigan, Illinois, and Wisconsin have documented their efforts or intentions to develop and involve advisory committees both in agenda setting and policy formation as well as during implementation, review, and evaluation. The engagement for the Department of Health and Human Services was structured to provide policy guidance and was operating primarily at the agenda-setting, analysis, and policy-formation stages but not in implementation, review, and evaluation.

A variety of approaches were used to engage stakeholders, although a key method was exchanges through advisory committees. Noting that in the Michigan program, initial engagement methods included focus groups and pre and posttest surveys assessing community attitudes is important. As a result of these engagement methods, a BioTrust Community Values Board was developed and has been implemented. This board advises the Michigan Department of Community Health on BioTrust program policies and procedures.

Finally, the greatest diversity across the NBS engagement examples was with respect to the different policy outcomes. The Michigan and Wisconsin engagements reported resulting

guidance on key issues facing each program (consent and data access for Michigan and evidentiary decision making for adding new disorders to the screening panel). The Illinois example is complicated; the advisory committee's role was to provide input on whether to add additional tests to the screening panel. However, an independent parent advocacy organization lobbied the legislature to add five lysosomal disorders to the screening panel that were not currently approved by the NBS program or the advisory committee. This example is important for two reasons. First, it points to the fact that there are many exogenous factors that influence the policy-development process. Second, it highlights the fact that engagement of a single stakeholder (such as an advisory board) may not be sufficient to represent all the important stakeholders for a particular issue.

Engagement examples: similarities and differences

Stakeholder engagement is widely lauded as a tool for improving clinical, scientific, and public health policy decision making. The two broad genomics examples examined support the fact that organizations (from state public health departments (in the case of NBS) to individual biorepositories) have an interest in engaging communities and stakeholders. The stakeholders involved, the purpose of the engagement, the policy-development stage(s), the methods used, and the outcomes varied greatly *between* the two contexts. *Within* each of the two broad areas, however, there were similarities in the types of stakeholders involved and engagement methods used. For example, within the biobanking context, stakeholders often were defined as biobank participants or members of the public; the engagement was often a multipronged approach with both one-time engagements and ongoing advisory groups to inform the development of biobanking policies, such as data sharing, or to advise on new and ongoing biobanking procedures. By contrast, stakeholders in the NBS examples were typically defined as disease advocates, parents of affected children, advocacy organizations, scientific and medical experts, and public health professionals. Information from the engagements sometimes were used to inform state and national policies and recommendations, to develop new consent procedures for families in the context of the use of leftover bloodspots in research, and to develop procedures for considering how to weigh evidence and determine which tests should or should not be added to the NBS panel.

Interestingly, in both contexts, at least half of the examples involved engagements that occurred throughout all stages of the policy-development cycle. This is likely because many of the engagements involved the use or development of an advisory committee or board. While an advisory committee provides a mechanism for gathering stakeholder feedback throughout policy development, how and whether the feedback is used to inform genomic policies is not clear.

Furthermore, one finding across the two genomics examples was that there were no reports of formal evaluations of the stakeholder engagement process related to policy development. To date, we have been unable to find any reported evaluations of how genomics policy decision makers assess key stakeholder input or details about how engagement findings are incorporated into genomics policy decisions or ongoing policy evaluation.

DISCUSSION

Seeking and understanding key stakeholder perspectives is recognized as an important component of developing sound public health, clinical, and research policies. Human genomics studies are increasingly including stakeholder and community engagements to elicit broader views on emerging areas, and many ethical, legal, and social issues have been explored through funded studies. Findings from these studies may increase our understanding of key stakeholder perspectives; however, whether and how these outcomes are used to inform policies is not well known. Engagements conducted specifically to inform new or existing genomics policies are lacking in the literature. Therefore, we chose to focus on two contexts in which there were clear, documented examples of stakeholder engagement in policy development.

Genomics is in a state of rapid technological change, and these technologies often trigger new policy decisions to address new knowledge. However, stakeholder engagement for genomics policy development is still at a nascent stage. Clearly, there are a variety of goals for any given stakeholder engagement, and each undertaking is context-dependent. In addition, some policies are more controversial and time-dependent than others. Those involving public legislation, or those that affect all members of society, may also require broader stakeholder input. Several different challenges of using stakeholder engagement as a tool for genomics policy development remain, and little evidence is currently available regarding how best to incorporate stakeholder feedback into policy-making processes.

Clinical policy development, such as policies guiding specific genetic/genomic testing or clinical genomic technologies, is absent from our case examples because stakeholder engagement is still in its early stages in this area of policy development, and there were few documented examples. There are several possible reasons for this difference. For example, there are different time frames for decision making in the clinical, public health, and research contexts. For example, biorepositories often unfold over a fairly long time frame, including securing funding, recruiting participants, obtaining samples, and sharing data with researchers. This provides a structure for stakeholder engagement over several years for a single biorepository. Furthermore, while there may be time-sensitive decisions and the need for adaptive governance structures to respond to new technologies and research priorities, there is an opportunity to establish stakeholder engagement up front. By contrast, clinical genomics policies are often time-sensitive because new technologies may be adopted before evidence-based policies are established. This underscores the need for rapid, robust stakeholder engagement processes. There are also significant structural differences between how policy is developed and who develops it across each of the three contexts. For example, NBS is a public health program, and policies must involve and adhere to statutory mandates. By contrast, a variety of organizational entities are involved in the development and implementation of clinical genomics policies, and the processes used in guideline development vary in how knowledge and evidence are synthesized.[34] Finally, identifying the appropriate stakeholders for any genomics policy decision is challenging. In general, we know very little about how key stakeholders are selected and how their input is used in clinical genomics policy development. Given that several instances of challenges in including, or not including, key stakeholders have been described in the formulation of

clinical genomics guidelines and recommendations,[35–41] this clearly is an area in need of greater research and development.

While this article is premised on the utility of stakeholder engagement, acknowledging that there are also important limitations and unintended consequences of engaging stakeholders is important. Stakeholders have different stated and unstated biases and interests that may sway policy decisions in a direction that is not necessarily based on scientific or clinical evidence. For example, the involvement of advocacy groups in NBS policy development could identify the expectations or concerns of constituencies dealing with a particular genetic disease, which in turn could influence legislatures to add a condition to a NBS panel without appropriate scientific evidence. This is reflected in the Illinois example in Table 2, where a parent advocacy group was successful in adding additional disorders to the panel in advance of having sufficient data on effective therapies and when treatment is necessary. This is not to suggest that stakeholder perspectives should not be incorporated or solicited, but rather that a single stakeholder group cannot represent all perspectives of the issue. Feedback from multiple stakeholders (including medical and scientific experts) should be weighed with scientific evidence, cost/benefit data, and other important factors. Furthermore, each of these types of evidence and perspectives may be valued or used differently in policy decision making depending on the context, as discussed above.

FUTURE PRIORITIES

Empirical research and conceptual work are needed to identify and define best practices for involving key stakeholders in different genomics policy-making contexts. Research efforts to explore when and in which contexts engagement is most helpful in policy development and how to conduct engagements in a cost-effective manner will help guide future practice. There is also a need for valid and reliable tools for systematically assessing and reporting the quality of stakeholder engagement and the policy process. Specifically, most of our case examples reported the use of community advisory boards and panels as a method for engaging community members and stakeholders across a variety of different programmatic and policy decisions. How the community advisory boards were constituted or moderated and how their input was integrated into policy and programmatic decisions were not clear. Future research should focus on establishing a framework for evaluating the quality and process of community advisory boards, and other engagement methods, to determine which approaches are most appropriate for a specific policy decision-making context or question.

Assessing the views of key stakeholders on their expectations for outcomes of engagements also will be important in developing future outcome measures. Very few data are available regarding potential barriers to and facilitators for integration of stakeholder input into genomics policy decision making—another area well suited for research. Further study in these areas will be valuable to inform first steps in the development of guidance principles or best practices for conducting effective stakeholder engagement in diverse genomics policy-making arenas.

In summary, there seems to be interest in and support for stakeholder engagements in a variety of areas in genomics policy making. Evidence-based best practices for conducting

engagements to inform the genomics policy process are, however, lacking. Those involved in genomics policy development should be encouraged to pursue stakeholder engagement, adapting it to their particular contexts, and to document stated goals, methods, and evaluation of their policy-informing processes. Ongoing research is needed to clarify rationales and potential outcomes, understand the perspectives of different stakeholders, and evaluate different approaches in genomics policy making. To prioritize this research agenda, possibly key stakeholder engagements could be used to focus future work in this area.

ACKNOWLEDGMENTS

The authors acknowledge the following members of the Trans-CEER Stakeholder Engagement Working Group for their valuable input on an earlier draft of this article: Wylie Burke, Dana Gold, Barbara Koenig, Carol Somkin, and Joon-Ho Yu. This work was funded by the National Human Genome Research Institute (P50 HG 003374, P20 HG 007243-01, and R21 HS023547).

REFERENCES

1. Graham, R.; Mancher, M.; Wolman, DM.; Greenfield, S.; Steinberg, E. Clinical Practice Guidelines We Can Trust. Washington, DC: National Academies Press; 2011.
2. Burton H, Adams M, Bunton R, Schröder-Bäck P. Developing stakeholder involvement for introducing public health genomics into public policy. Public Health Genomics. 2009; 12:11–19. [PubMed: 19023186]
3. Callaway E. Deal done over HeLa cell line. Nature. 2013; 500:132–133. [PubMed: 23925220]
4. Sniderman AD, Furberg CD. Why guideline-making requires reform. JAMA. 2009; 301:429–431. [PubMed: 19176446]
5. Terry SF. Don't just invite us to the table: authentic community engagement. Genet Test Mol Biomarkers. 2013; 17:443–445. [PubMed: 23721344]
6. Terry SF, Bonhomme N. Nothing about us without us: guidelines for genetic testing. Genet Test Mol Biomarkers. 2013; 17:357–358. [PubMed: 23611249]
7. Barry MJ, Edgman-Levitan S. Shared decision making–pinnacle of patient-centered care. N Engl J Med. 2012; 366:780–781. [PubMed: 22375967]
8. Carman KL, Dardess P, Maurer M, et al. Patient and family engagement: a framework for understanding the elements and developing interventions and policies. Health Aff (Millwood). 2013; 32:223–231. [PubMed: 23381514]
9. Fleurence R, Selby JV, Odom-Walker K, et al. How the Patient-Centered Outcomes Research Institute is engaging patients and others in shaping its research agenda. Health Aff (Millwood). 2013; 32:393–400. [PubMed: 23381533]
10. Hoffman A, Montgomery R, Aubry W, Tunis SR. How best to engage patients, doctors, and other stakeholders in designing comparative effectiveness studies. Health Aff (Millwood). 2010; 29:1834–1841. [PubMed: 20921483]
11. Krahn M, Naglie G. The next step in guideline development: incorporating patient preferences. JAMA. 2008; 300:436–438. [PubMed: 18647988]
12. O'Haire, C.; McPheeters, M.; Nakamoto, E., et al. Engaging Stakeholders to Identify and Prioritize Future Research Needs. Rockville, MD: Department of Health and Human Services. Agency for Healthcare Research and Quality; 2011.
13. Puddy, RW.; Wilkins, N. Understanding Evidence Part 1: Best Available Research Evidence. A Guide to the Continuum of Evidence of Effectiveness. Atlanta, GA: Centers for Disease Control and Prevention; 2011.
14. Principles of Community Engagement. 2nd edition. Bethesda, MD: Department of Health and Human Services. National Institutes of Health; 2011. CTSA Community Engagement Key Function Committee Task Force on the Principles of Community Engagement.
15. International Association for Public Participation (IAP2). IAP2's Public Participation Toolbox. 2006 http://c.ymcdn.com/sites/www.iap2.org/resource/resmgr/imported/06Dec_Toolbox.pdf.

16. Hurle B, Citrin T, Jenkins JF, et al. What does it mean to be genomically literate?: National Human Genome Research Institute Meeting Report. Genet Med. 2013; 15:658–663. [PubMed: 23448722]
17. O'Doherty KC, MacKenzie MK, Badulescu D, Burgess MM. Explosives, genomics, and the environment: conducting public deliberation on topics of complex science and social controversy. SAGE Open. 2013; 3:1–17.
18. Syurina EV, Brankovic I, Probst-Hensch N, Brand A. Genome-based health literacy: a new challenge for public health genomics. Public Health Genomics. 2011; 14:201–210. [PubMed: 21734434]
19. Mello MM, Wolf LE. The Havasupai Indian tribe case–lessons for research involving stored biologic samples. N Engl J Med. 2010; 363:204–207. [PubMed: 20538622]
20. Ramshaw E. DSHS turned over hundreds of DNA samples to Feds. Texas Tribune. 2010 http://www.texastribune.org/2010/02/22/dshs-turned-over-hundreds-of-dna-samples-to-feds/.
21. Carmichael M. Newborn screening: a spot of trouble. Nature. 2011; 475:156–158. [PubMed: 21753828]
22. Beleno v Texas Dept. of State Health Services. et al. Case No. 5:09-cv-00188. US District Court for the Western District of Texas in San Antonio; 2009.
23. MN Supreme Court rules on newborn screening samples. Minn Med. 2011; 94:18–19.
24. Callaway E. NIH director explains HeLa agreement. Nature News. 2013 http://www.nature.com/news/nih-director-explains-hela-agreement-1.13521.
25. Caplan A. NIH finally makes good with Henrietta Lacks' family—and it's about time, ethicist says. 2013. NBC News. 2013 Aug 7. http://www.cnbc.com/id/100946766.
26. Coveney J. Analyzing public health policy: three approaches. Health Promot Pract. 2010; 11:515–521. [PubMed: 18515504]
27. Bridgman, P.; Davis, G. The Australian Policy Handbook. 3rd ed.. Crows Nest, NSW, Australia: Allen & Unwin; 2004.
28. Henderson GE, Cadigan RJ, Edwards TP, et al. Characterizing biobank organizations in the U.S.: results from a national survey. Genome Med. 2013; 5:3. [PubMed: 23351549]
29. Haldeman KM, Cadigan RJ, Davis A, et al. Community engagement in US biobanking: multiplicity of meaning and method. Public Health Genomics. 2014; 17:84–94. [PubMed: 24556734]
30. American College of Medical Genetics and Genomics. Newborn screening: toward a uniform screening panel and system. Genet Med. 2006; 8(suppl 1):1S–252S. [PubMed: 16783161]
31. American Academy of Pediatrics. Serving the family from birth to the medical home. Newborn screening: a blueprint for the future - a call for a national agenda on state newborn screening programs. Pediatrics. 2000; 106(2 Pt 2):389–422. [PubMed: 10947682]
32. Olson, S.; Berger, AC. Challenges and Opportunities in Using Residual Newborn Screening Samples for Translational Research: Workshop Summary. Washington, DC: National Academies Press; 2010. Institute of Medicine (US) Roundtable on Translating Genomic-Based Research for Health.
33. Therrell BL Jr, Hannon WH, Bailey DB Jr, et al. Committee report: Considerations and recommendations for national guidance regarding the retention and use of residual dried blood spot specimens after newborn screening. Genet Med. 2011; 13:621–624. [PubMed: 21602691]
34. Schully SD, Lam TK, Dotson WD, et al. Evidence synthesis and guideline development in genomic medicine: current status and future prospects. Genet Med. 2015; 17:63–67. [PubMed: 24946156]
35. Yu JH, Harrell TM, Jamal SM, Tabor HK, Bamshad MJ. Attitudes of genetics professionals toward the return of incidental results from exome and whole-genome sequencing. Am J Hum Genet. 2014; 95:77–84. [PubMed: 24975944]
36. Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group. The EGAPP initiative: lessons learned. Genet Med. 2014; 16:217–224. [PubMed: 23928914]
37. Burke W, Antommaria AH, Bennett R, et al. Recommendations for returning genomic incidental findings? We need to talk! Genet Med. 2013; 15:854–859. [PubMed: 23907645]

38. Ross LF, Rothstein MA, Clayton EW. Mandatory extended searches in all genome sequencing: “incidental findings,” patient autonomy, and shared decision making. JAMA. 2013; 310:367–368. [PubMed: 23917281]
39. Ross LF, Rothstein MA, Clayton EW. Premature guidance about whole-genome sequencing. Per Med. 2013; 10
40. Allyse M, Michie M. Not-so-incidental findings: the ACMG recommendations on the reporting of incidental findings in clinical whole genome and whole exome sequencing. Trends Biotechnol. 2013; 31:439–441. [PubMed: 23664778]
41. Townsend A, Adam S, Birch PH, Friedman JM. Paternalism and the ACMG recommendations on genomic incidental findings: patients seen but not heard. Genet Med. 2013; 15:751–752. [PubMed: 24008256]
42. Lemke AA, Wu JT, Waudby C, Pulley J, Somkin CP, Trinidad SB. Community engagement in biobanking: Experiences from the eMERGE Network. Genomics Soc Policy. 2010; 6:35–52. [PubMed: 22962560]
43. National Cancer Institute, National Institutes of Health, Office of Biorepositories and Bioscpecimen Research. [Accessed 6 March, 2014] Summary: National Cancer Institute Biospecimen Best Practices Forum. 2007. http://biospecimens.cancer.gov/practices/forum/boston2007/pdf/FINAL_11-05-07_NCI_BPs_Forum_Boston_Summary_Rev1-24-08_Ed.pdf.
44. National Institutes of Health (NIH). [Accessed 6 March, 2014] Request for Information: Input on the Draft NIH Genomic Data Sharing Policy. 2013. http://grants.nih.gov/grants/guide/notice-files/NOT-OD-13-119.html.
45. National Institutes of Health, Department of Health and Human Services. Policy for sharing of data obtained in NIH supported or conducted genome-wide association studies (GWAS). 72 FR 49290. E7-17030. 2007; |29290–49297.
46. O’Doherty K, Ibrahim T, Hawkins A, Burgess M, Watson P. Managing the introduction of biobanks to potential participants: lessons from a deliberative public forum. Biopreserv Biobank. 2012; 10:12–21. [PubMed: 24849749]
47. Rotimi C, Leppert M, Matsuda I, et al. International HapMap Consortium. Community engagement and informed consent in the International HapMap project. Community Genet. 2007; 10:186–198. [PubMed: 17575464]
48. Duquette D, Langbo C, Bach J, Kleyn M. Michigan BioTrust for Health: public support for using residual dried blood spot samples for health research. Public Health Genomics. 2012; 15:146–155. [PubMed: 22488457]
49. Ross LF, Waggoner DJ. Parents: critical stakeholders in expanding newborn screening. J Pediatr. 2012; 161:385–389. [PubMed: 22727864]
50. [Accessed 6 March, 2014] Charter for Illinois Genetic and Metabolic Advisory Committee. http://www.ilga.gov/legislation/ilcs/ilcs3.asp?ActID=2956&ChapterID=35.
51. Newborn Screening Task Force. Report to the Secretary of the Wisconsin Department of Health Services. 2013
52. Secretary of Health and Human Services. Charter: Discretionary Advisory Committee on Heritable Disorders in Newborns and Children. Washington, DC: Department of Health and Human Services; 2013.
53. Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, Department of Health and Human Services. [Accessed 6 March, 2014] Recommended uniform screening panel. 2013. http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/index.html.

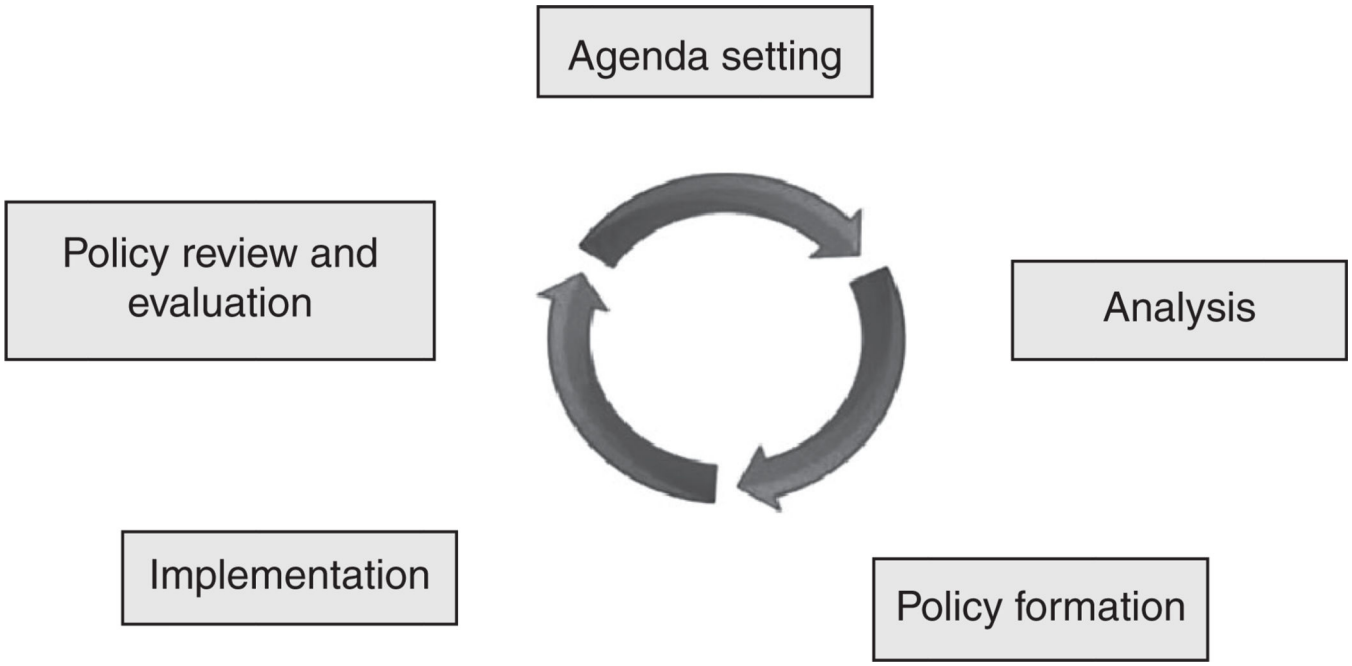

Figure 1.
Stakeholder engagement at various stages of the policy-making cycle.

Table 1

Reasons to engage stakeholders in genomics policy issues

1. To identify areas of commonality/agreement
2. To Identify areas of disagreement and determine what is driving those differences
3. To help articulate/reflect values of the broader community that is affected
4. To align practice recommendations with societal needs and expectations
5. To improve overall genetic literacy and understanding of mutual perspectives
6. To help execute the implementation of guidelines as intended
7. To promote transparency
8. To increase the quality and trustworthiness of the policy

Table 2

Examples of stakeholder engagements used to inform biobank research policies

Institution or organization	Key stakeholders	Purpose of the engagement related to policy	Policy-development stage	Methods of engagement	Outcomes of engagement related to policy
KP RPGEH[42]	KP and community members, participants, refusers, investigators, IRB members, CAP, funders, KP leadership	To develop biobank recommendations and best practices and disseminate results and recommendations	Agenda setting, analysis, policy formation, implementation, policy review and evaluation	Focus groups, mail surveys, telephone interviews, CAP	CAP recommendation that RPGEH use an opt-in mechanism for reconsent was adopted
NIH NCI, Office of Biorepositories and Biospecimen Research[43]	Intramural and extramural cancer researchers, patient communities	To inform, update, and plan for future versions of NCI best practices	Agenda setting, analysis, policy formation, implementation, policy review and evaluation	Surveys, community forums, symposiums, webcasts, public comments	Establishment of a Biorepository Coordinating Committee, development of guidelines and best practices for NCI-supported biorepositories
Mayo Clinic[42]	Minnesota residents, biobank participants, researchers, clinicians, Mayo Clinic leadership, ethicists, IRBprofessionals	To develop recommendations for biobank procedures and policies; obtain ongoing input and guidance on biobank operations	Agenda setting, analysis, policy formation, implementation, policy review and evaluation	Deliberative democracy, mail surveys, in-person interviews, observation of consent process, CAB	Development and use of a simplified informed consent process; an option for participants to withdraw from the biobank; creation of a CAB
NIH[44,45]	US public, genomic researchers, research participants	To inform NIH policy for sharing of data obtained in NIH-supported or conducted GWASs and policy for genomic data sharing	Policy formation, policy review and evaluation	Town hall meetings, online policy draft for public comment, public consultation webinar	GWAS policy finalized in 2007; Genomic Data Sharing policy drafted in 2013; public comments posted
BC BioLibrary[46]	Diverse British Columbia residents	To inform specific protocols and governance structures of the BC BioLibrary	Agenda setting, policy formation	Deliberative democracy	Implemented “permission to contact processes” for potential donors to be contacted by a representative of a biobank to discuss biobank research participation
International HapMap Project[47]	Nigerian, Japanese, Chinese, and US (Utah) residents	To provide input into how the samples from their locality would be collected and described	Policy formation, implementation	Interviews, focus groups, surveys, public meetings, personal visits, community advisory groups	Labeling samples to reflect social identities; Statement of Research Intent modified to use lay language

CAP, community advisory panel; GWAS, genome-wide association study; IRB, institutional review board; KP, Kaiser Permanente; NCI, National Cancer Institute; NIH, National Institutes of Health; RPGEH, Research Program on Genes, Environment and Health.

Table 3

Examples of stakeholder engagements used to inform newborn screening policies

Institution or organization	Key stakeholders	Purpose of the engagement related to policy	Policy-development stage	Methods of engagement	Outcomes of engagement related to policy
Michigan's BioTrust for Health/ Michigan Department of Community Health[48]	Ten diverse communities selected to represent special concerns as stakeholders in the BioTrust	To ensure citizen involvement in the development of policies governing the use of stored dried blood spots from newborn screening	Agenda setting, analysis, policy formation, implementation, review and evaluation	Focus groups; pre- and postsurveys; BioTrust Community Values Board	Retained an opt-out policy for the use of archived bloodspots and formalized an opt-in policy for new leftover bloodspots; developed research guidelines and a multistage review process
Illinois Department of Public Health[49,50]	Appointed members of the Genetic and Metabolic Advisory Committee (parents of affected children, scientific experts, medical experts)	To evaluate the appropriateness of adding conditions to the NBS panel and advise on all aspects of the NBS program	Agenda setting, analysis, policy formation, implementation, review and evaluation	Genetic and Metabolic Advisory Committee meetings	The Evanosky Foundation (parent advocacy group) lobbied the legislature to mandate screening for additional lysosomal storage diseases in Illinois' NBS program
Wisconsin NBS Program[51]	Wisconsin NBS Task Force (parents of affected children, physicians, public health experts), Wisconsin NBS Umbrella Committee (parent representatives, March of Dimes, Hospital Association)	To propose a framework for making addition and deletion decisions by scientifically weighing evidence and eliminating bias	Agenda setting, analysis, policy formation, implementation, review and evaluation	Meetings and communications of the Wisconsin NBS Task Force, Wisconsin NBS Umbrella Committee	Provided recommendations on how to improve the advisory process and developed criteria for adding/deleting tests
US Department of Health and Human Services (Federal NBS Policy)[52]	Members of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (medical, technical, public health, or scientific professionals; experts in ethics and heritable disorders; patient advocates	To provide advice to the Secretary of Health and Human Services about aspects of NBS and childhood screening and technical information for the development of policies and priorities in NBS	Agenda setting, analysis, policy formation	Meetings and communications of the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children	Provided recommendations for national policy on retention and use of dried blood spot specimens after NBS; developed a decision-making process for including conditions in Recommended Uniform Screening Panel[53]

NBS, newborn screening.