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The Ethical Challenges of Newborn Screening Programs in the United States

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THE ETHICAL CHALLENGES OF NEWBORN SCREENING PROGRAMS IN THE UNITED STATES

By

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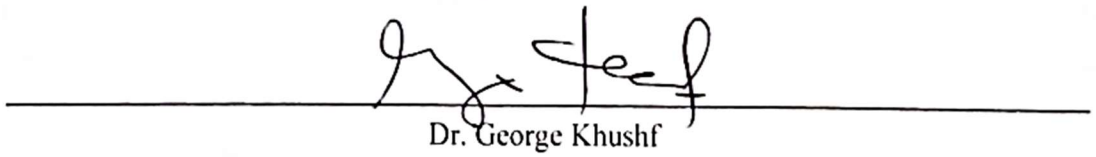

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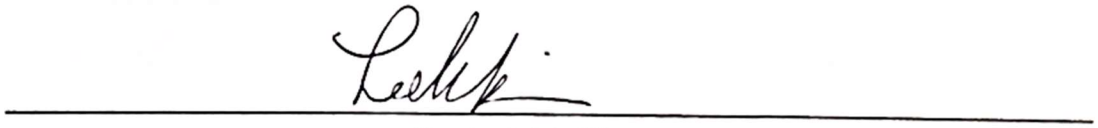

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Thesis Summary

Newborn screening programs have been mandated throughout the United States since the 1960s, and technological advancements have allowed for their evolution into the essential public health entities they are today (Arnold 558). These programs screen newborns for a variety of congenital and genetic conditions in all states, but each state varies in conditions screened and policies for collecting and using samples. Residual blood spots are a key component of these programs because they are often used for secondary purposes, such as for quality assurance and public health or biomedical research (Botkin et al. 121). Ethical challenges have arisen related to consent and privacy policies employed by states. Legal cases have arisen surrounding the accessibility of these blood spots by outside entities, such as law enforcement agencies. Problems with informed consent, privacy protections, and transparency within many states' programs call for the alteration of policies so that citizens' autonomy and privacy are upheld and so that trust and support for these public health initiatives are sustained.

Introduction

Since the 1960s, newborn screening has been utilized throughout the United States (Arnold 558). Testing for phenylketonuria (PKU) via bacterial inhibition assay was developed by Robert Guthrie, and this became the first stable metabolic test implemented on a wide scale for newborns (Ross 300). PKU is a metabolic disorder that results in the accumulation of an amino acid within the body, resulting in brain damage if dietary changes are not made within the first few months of life (Ross 300). The test for PKU involves the quantification of molecules within dried blood spots from newborns, a technique that remains in use today but at a much larger scale (Caggana et al. 15). Mandated testing for PKU was begun and is still used as a way to detect the disease and implement early treatment to minimize detrimental effects (Caggana et al. 15).

The implementation of tandem mass spectrometry during the 1990s was instrumental in the expansion of newborn screening programs because this method allows for the quantification of many metabolites within a small amount of blood (Elster 181). Compared to previous metabolic tests, such as the one that screens for PKU, much less time and resources are required to test samples for a variety of disease markers using this method (Ross 309). These advances in instrumental methods have allowed for the expansion of testing to an array of disorders, including sickle cell anemia and hypothyroidism (Tarini and Goldenberg 770). Additionally, testing methods have expanded to include the testing of DNA, allowing for the detection of genetic disorders like cystic fibrosis (Caggana et al. 16). Multiplex testing via tandem mass spectrometry alone has created a tremendous pool of individuals with conditions that could be further understood through their utilization in research, and the inclusion of genetic testing has amplified this outcome (Ross 309).

Residual Blood Spots

All states currently mandate screening of a variety of conditions for all newborns (Rothwell et al., "An Assessment" 335). These tests are conducted via a heel prick, typically shortly after birth and again within a few weeks of age (Bick et al.). After testing is initially performed on blood samples collected from newborns, the residual blood spots are stored and possibly used for secondary purposes, depending on state guidelines (Caggana et al. 17).

The storage of residual blood spots is somewhat analogous to biobanks, which are institutions that retrieve biospecimens from donors and perform research on them (Knoppers et al. 4). Biobanks primarily aim to identify and treat conditions within the population, but they also function as a major contributor to biomedical research, providing institutions with the data needed to establish links between genotype and disease (Knoppers et al. 4). Subtypes include population biobanks that focus on longitudinal research, disease biobanks which are typically tied to clinical purposes, and residual tissue biobanks (Knoppers et al. 4).

Residual blood spots are stable for long periods of time, so they can be stored indefinitely and have data extracted from them at any given time (Botkin et al. 121). These stored samples are commonly used for quality assurance purposes (Botkin et al. 121). This practice of regularly evaluating the accuracy of methods already in place is necessary to validate results for all analytical methods, and this use does not involve the transfer of blood spots to outside entities. Samples may also be used for secondary purposes, such as research and forensic uses, which may involve the transfer of data or physical specimens between institutions (Botkin et al. 121).

The Justification of Mandated Newborn Screening

The Wilson and Jungner criteria were developed in 1968 in order to evaluate the potential of conditions considered for screening (Dobrow et al. E422). These principles claim that the condition should be a health problem that is well understood with an established treatment plan (Dobrow et al. E423). This set of guidelines has been the traditional approach to determining the conditions included in newborn screening programs, but potential expansions may involve a departure from these criteria (Downie et al.). Advancements in methods have reduced the amount of resources required to test for many conditions, leading to the expansion in conditions screened over time. Further developments, especially within genomics, could impact newborn screening in the near future. Despite these potential shifts in newborn screening criteria, consent and privacy practices within states have remained largely unchanged.

States have justified mandated screening programs through parens patriae, a principle which asserts that the state can override parental authority if the goal is to protect a child's health (Zacharias et al. S39). Historical uses of other police powers by states include mandated quarantines and vaccines, as seen with the COVID-19 pandemic, and other public health initiatives such as restrictions on public smoking and prescription use (Whelan 435). This emphasis on advancing public health is a central argument for the absence of informed consent processes and the loose privacy protections in many states' newborn screening procedures.

Potential Expansions

Traditional approaches to newborn screening are based on the identification of conditions through metabolic screening, which does not require the analysis of genomic sequences (Downie et al.). The expansion of screening programs to include genetic testing would allow for the

detection of additional conditions that may not have been found using traditional methods. Some genomic newborn screening programs test hundreds of genes, effectively screening for multiple conditions at many areas of the genome (Downie et al.).

Whole genome sequencing (WGS) is a technique that could be employed for such testing in the future, where the entire genetic sequence is determined and then areas of interest are analyzed (Bick et al.). This approach could also be utilized to determine the risk of a newborn developing health problems later in life, such as certain types of cancers or autoimmune diseases (Bick et al.). In addition to the ethical challenges associated with current programs, this potential expansion could amplify these concerns and pose a larger threat to the maintenance of public trust. Genomic screening methods may expand opportunities for outside entities to extract genetic information from stored samples, igniting concerns about obtaining true informed consent from parents and the overall accessibility of samples.

A Survey of Newborn Screening Practices in the United States

State-mandated programs have evolved along with testing methods, producing a variety of approaches for obtaining samples from newborns, the extent of testing these samples undergo, and what happens to the samples after testing (Caggana et al. 15). Concerns regarding the ethics of such programs have arisen over time, especially regarding parental consent and access to samples by other institutions.

Informed consent practices throughout the country range from a complete absence in some states to thorough procedures that highly involve parents in others. Additionally, some established informed consent processes involve informing parents of possible secondary uses of residual blood spots while others do not (Lewis et al. 707). Policies regarding the permitted

secondary uses for samples collected from newborns also vary among states, with some that highly restrict access to a clearly defined set of institutions and others that allow extensive uses, such as for research or forensic purposes (Botkin et al.). The absence of informed consent practices in some states and the use of newborn blood samples for secondary reasons unknown to parents demonstrate a deviation from traditional medical ethical principles, demonstrating potential tension between public health and these traditional principles.

Legal cases have resulted from states' practices regarding newborn screening, with *Beleno et al. v. Texas Department of State Health Services* being one of the most notable. In this case, the plaintiffs claimed that their consent was not obtained for secondary research performed with their children's samples nor were they notified of such uses (Lewis et al. 704). A settlement was reached between the parties, resulting in the destruction of millions of blood spots within the state (Lewis et al. 704). The outcome of this case involved the loss of a major resource for biomedical research and may have negatively impacted the public health practice of monitoring and mitigating disease in newborns, yet it also demonstrates the presence of concerns within the general public regarding the state's respect for their autonomy and privacy. This case is one of many that has raised questions about newborn screening practices within the nation.

Two representative cases involving ethical concerns about informed consent processes and the secondary uses of stored residual blood spots illustrate the extremes that encircle the variability in newborn screening programs throughout the United States. The outcome of a case in Minnesota led to the construction of consent practices that involve parents at multiple steps in the process as well as the restriction of uses for blood spots within the state (Ram 1264). Conversely, an ongoing case in New Jersey demonstrates concerns associated with a state-mandated program that lacks informed consent processes and strong restrictions surrounding the

use of blood spots (Superior Court of New Jersey). Considering these cases together allows for the assessment of variable state policies as well as the arguments made for their justification.

Newborn Screening in Minnesota

A 2011 case, *Bearder v. State*, centered around nine families who sued the state of Minnesota for neglecting to obtain informed consent from parents before collecting samples from their newborns, storing them for extended periods of time, and sharing them with research institutions (Minnesota Supreme Court). The plaintiffs claimed that these practices were in direct violation of the Genetic Privacy Act outlined in the Minnesota Government Data Practices Act, which defines protections for certain types of genetic information (Minnesota Supreme Court). Because of this, the court had to determine if newborn screening samples were classified as genetic information and if the Genetic Privacy Act applied to programs that collected them, which would imply the overriding of previously enacted statutes (Minnesota Supreme Court).

The district court of Minnesota initially ruled that the blood samples obtained from newborns were not classified as genetic information and that the newborn screening statutes previously established within the state apply regardless of the Genetic Privacy Act (Minnesota Supreme Court). Further complicating Minnesota's views of genetic data, the court of appeals ruled that newborn blood samples are indeed genetic information, yet the newborn screening statutes still take precedence over the Genetic Privacy Act (Minnesota Supreme Court). The court of appeals also stated that the use of these blood samples outside of screening processes violate the Genetic Privacy Act, but due to a lack of evidence of such practices, a ruling was not made regarding this opinion (Minnesota Supreme Court). This led to the case being brought to

the Supreme Court of the state, resulting in a ruling that dramatically altered the operation of newborn screening within Minnesota.

The Supreme Court reversed the original decision made by the court of appeals, citing ambiguous language present in the definition of genetic information provided by the Genetic Privacy Act as well as the restrictions outlined regarding the storage and use of samples (Minnesota Supreme Court). The court decided that blood samples from newborns themselves are considered genetic information since they are biological in nature and that the analysis of DNA from samples is not required to meet this definition (Minnesota Supreme Court). Additionally, it was ruled that the newborn screening statutes do hold some precedence over the Genetic Privacy Act regarding the collection and storage of newborn blood samples as they relate to testing for congenital and genetic disorders (Minnesota Supreme Court). However, these statutes do not express that blood samples can be used, stored, or shared for purposes outside of testing for these conditions (Minnesota Supreme Court).

This ruling enforced that informed consent is required for blood samples to be used and disseminated for secondary purposes like research (Minnesota Supreme Court). As a result, the Minnesota Department of Health was ordered to destroy any blood samples collected after the date of the decision, and the laws were updated to reflect the court's view on informed consent (Collins). The newborn screening statutes currently state that parents must be informed of newborn screening practices, including the type of testing initially performed on samples to possible other uses for program operations and research pertaining to public health initiatives (2022 Minnesota Statutes). Limited definitions of program operations and public health research are provided in the legislation, asserting that no other tasks can be carried out using blood samples unless written consent is obtained from the parents (Ram 1264). This bars access to

newborn blood samples from many outside institutions, including law enforcement, which is an approach that differs from other states (Ram 1284). The statutes also include a clause that permits parents to withdraw consent in regard to the storage and use of collected samples from their children, which would result in their destruction (2022 Minnesota Statutes).

Bearder demonstrates the complex nature of newborn screening programs within a state, especially surrounding how definitions provided in legislation can affect the rights of parents and newborn screening programs as it applies to the storage, internal use, and sharing of samples. As a result of this case, Minnesota has adopted a stricter approach to newborn screening, requiring parents to provide written informed consent for the use of samples for certain secondary research (2022 Minnesota Statutes). Access to residual blood spots within Minnesota by outside institutions without consent is significantly more restricted than other states, demonstrating the complex framework of newborn screening programs throughout the nation (Ram 1284).

Newborn Screening in New Jersey

In New Jersey, an ongoing case, *NJ Office of the Public Defender et al. v. Department of Health et al.,* involves the access to stored residual blood spots by law enforcement agencies (Superior Court of New Jersey). Plaintiffs in this case claim that the State Police conducted an unlawful search by gaining access to a blood sample collected from a newborn nine years prior and using its genetic information to confirm that a client of the Office of the Public Defender (OPD), the child's father, committed a crime in 1996 (Superior Court of New Jersey). The OPD asserts that the State Police did not have the authority to use this blood sample due to the lack of a warrant for collecting a DNA sample from its client (Superior Court of New Jersey).

Further concerns are associated with this case, including the fact that newborn blood samples are stored for decades within the state and are accessible by outside organizations (Superior Court of New Jersey). The apparent loose regulations associated with the storage and use of residual blood spots within New Jersey have raised questions regarding the privacy of newborns and their family members, as well as the process of obtaining consent for the use of these samples for purposes beyond screening for disorders present at birth.

Within New Jersey, there is not a requirement for parents to be informed about the initial screening of their children as well as any additional actions performed with the samples collected from newborns (Superior Court of New Jersey). The absence of an informed consent process poses a threat to autonomy on its own, and the ability of outside organizations to obtain access to collected samples without consent threatens the privacy of newborns and their relatives that can be traced through genetic testing (Superior Court of New Jersey).

State Police were essentially able to bypass the need for probable cause before requesting a DNA sample from the suspect by issuing a subpoena upon the state's Newborn Screening Laboratory (Superior Court of New Jersey). Through the use of genetic information extracted from a residual blood spot, law enforcement was then able to establish probable cause for the collection of the suspect's DNA (Superior Court of New Jersey). The OPD asserts that the privacy of their client was violated through these actions and that the analysis of a relative's DNA should qualify as a search that requires a warrant (Superior Court of New Jersey).

Legal definitions of what constitutes a search by law enforcement as well as what information can be utilized from residual blood spots play a large and likely deciding role in this case. How New Jersey responds to the ruling could dramatically alter the operation of newborn screening programs within the state. However, in its current condition, the lack of an informed

consent process and the ability of an array of institutions to access residual blood spots poses many ethical concerns within the state.

Newborn Screening Among States

These cases from Minnesota and New Jersey illustrate the variability of newborn screening programs within the United States. Plaintiffs cited similar concerns in both cases, especially related to informed consent and the access to residual blood spots by other institutions. Though an array of policies pertaining to newborn screening exist throughout the United States, concerns related to informed consent and privacy seem to be ubiquitous. The expansion of programs to test for a growing number of conditions over the years and the ability to extract more information from samples has increased concerns for consent, privacy, and transparency (Arnold 560).

As demonstrated by the legal cases presented, consent practices for newborn screening are highly variable among states. Many states, such as New Jersey, have not implemented a consent process at all for its screening programs (Superior Court of New Jersey). Other states have implemented practices of broad consent regarding newborn screening, which indicate that the parents' initial decision to participate in newborn screening implies their consent to further actions done with the collected samples, including secondary research or law enforcement activities (Rothwell et al., "An Assessment" 335).

Scholars have criticized such approaches to consent because research may emerge after consent was initially obtained, resulting in the use of samples for purposes not known by participants (Rothwell et al., "An Assessment" 335). Additionally, vague language may be used when obtaining consent, such as "use for other research," which may not fully inform

participants of possible future actions performed with collected genetic information (Minnesota Supreme Court). Following *Bearder*, Minnesota has demonstrated that participants can be informed of the specifics of secondary research, as their statutes list out a limited number of uses for residual blood spots within this realm (2022 Minnesota Statutes). Minnesota's current practices also require written informed consent from parents to opt-in to additional research using their child's samples, showing that the inclusion of parental decision-making can be implemented statewide for newborn screening (Ram 1264).

The Process of Informed Consent

Ethical principles that institutions aim to follow, which form the foundation of informed consent among other concepts, were outlined in the Belmont Report in 1979. This is a set of guidelines compiled by the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research that are followed in order to ensure adequate protection of research participants (The Belmont Report). Despite this targeted purpose, these concepts are presented in a general way that allows for their application to procedures outside of research, including practices within newborn screening.

Many ethical concepts build upon respect for people's autonomy and providing protections when autonomy is compromised (Konnoth 1321). Autonomy refers to the ability of a person to consider their own opinions and make their own choices without obstruction from outside sources (The Belmont Report). Informed consent builds upon this principle and refers to the process of an individual making an informed decision to participate in a process, ideally based on the communication of the benefits and risks associated with the service from the provider (Grant). This process typically involves a document that outlines the details of a

procedure, though some approaches may involve implied or broad consent from participants (Corneli et al. 14). This tends to be the case for newborn screening programs, as most states have adopted an opt-out method that assumes consent from parents unless otherwise stated (Ross 313).

This concept is largely context-dependent, meaning that some environments may adopt abbreviated approaches to obtaining consent in accordance with the procedure to be performed and the participant pool. Newborn screening serves a widescale public health purpose that involves the collection of samples from almost all newborns born within the country (Caggana et al. 14). Each year, newborns with genetic conditions are identified and treated due to this testing, so arguments could be made that these tests provide protection that would not be present without them, therefore justifying state-mandated programs.

The use of police powers by states is most applicable to targeted testing that identifies diseases that require immediate treatment in order to prevent detrimental health effects (Zacharias et al. S39). Justification for expanded testing is less apparent, particularly in the absence of consent and transparency. Whole genome sequencing, for example, would involve the extraction of information that does not pertain to conditions that meet the traditional criteria of newborn screening (Downie et al.). The mandatory nature of newborn screening is largely justified through its use as a detection tool so that treatments can be implemented for genetic and metabolic disorders. The absence of the critical step of treatment for some tested disorders suggests that developments within mandated newborn screening programs may involve an overstepping in their scope of practice.

Factors that Impact Consent

The ability of individuals to elect to participate in research voluntarily, without outside factors influencing their decision, is also necessary in order to retain autonomy (The Belmont Report). Coercion through the disproportionate presentation of harms that are possible if an individual declines to participate violate this concept, but this ignites debates about what exactly constitutes coercion (The Belmont Report). Risks and benefits exist for every study, and their disclosure is crucial for informed decisions to be made, so researchers must carefully present this information in a way that minimizes bias and coercion. It is likely that inherent bias exists within institutions, as they desire to recruit participants to further studies, so problems with informed consent may arise from this aspect. Newborn screening programs are largely mandatory due to their vital role in public health, so tension exists between this purpose and the ability of parents to have a say in what samples are collected from their children.

The way in which information is presented during the consent process is important since participants' comprehension of procedures can easily be affected due to factors such as language and other events surrounding the process. Generally, people have been nudged to participate in procedures during the presentation of the methodology, such as through the selective mentioning of positive outcomes and a minimal discussion of risks (Mehlman et al. 3). Some scholars have argued that nudges are built into opt-out consent models since participants may be less willing to withdraw from a process when participation is presented as the default option (Mehlman et al. 2).

Others may view the use of nudges as an effective way to strengthen participation in a procedure that advances public health, such as newborn screening programs. Previous studies have demonstrated that individuals are largely affected by biases, meaning that their decisions may not always be made in a rational manner (Vos et al. 735). The average American adult will

not achieve full understanding of the complexities of biomedical procedures despite the informed consent process, so there are questions regarding the effectiveness of such approaches. Participants' notions of the healthcare system may also play a role in the filtering of information provided by professionals, resulting in a skewed decision that does not truly align with the methodology communicated.

The use of language that stresses widespread participation may also pressure individuals to partake in a study without fully understanding the risks associated with it (Corneli et al. 21). For example, the presentation of the options themselves may affect the decision made by parents, so there must be careful consideration put into every part of a consent form to avoid bias. Certain approaches may utilize a list of options that present a variety of "yes" options and a single "no" option, which could nudge parents towards providing consent (Mehlman et al. 2) .

Healthcare professionals are more knowledgeable about the methodology of newborn screening and will likely influence participants based on this fact alone. Minimizing the disclosure of details about risks and altering consent methods to enhance participation rates may take advantage of the population's trust in healthcare. For instance, the omission or understatement of certain risks of screening programs could impact the decision ultimately made by parents (Mehlman et al. 1). If negative impacts of a study were to gain public attention as a result of such practices, public trust would be threatened and the population's cooperation with healthcare could be hindered.

Researchers have proposed that informed consent processes should focus on deliberating the risks and benefits of a procedure based on previous experiences and factual evidence in order to arrive at a rational decision (Vos et al. 737). The process of informed consent for newborn screening is absent in many states and abbreviated in others, raising concerns about the true

rationale behind parental decisions. The benefit to public health that newborn screening provides is often used to justify this less engaged approach to consent, yet these programs may impact areas outside of this scope.

Perhaps nudging parents through the careful presentation of newborn screening's vital role in public health is necessary to retain participation and minimize health-related risks for individuals. Nonetheless, the impact of misinformation and a lack of knowledge present within the population without informed consent poses a significant threat to the maintenance of public trust and support in these programs. Practicing respect for parental autonomy does not necessarily indicate that biases in the population will have a negative impact on participation as long as information is carefully and fully presented by providers.

Current Consent Processes

Past studies have determined that a majority of mothers do not possess knowledge about newborn screening or the associated storage and use of residual blood spots (Newcomb et al. 290). Current practices for informing parents about screening procedures occur during a stressful fast-paced time near birth, suggesting that parents may not fully grasp the newborn screening process when it is presented (Hasegawa et al. 301). This process may or may not involve obtaining explicit consent from the parents, as many states have implemented mandatory screening programs that operate under opt-out methodologies (Currier).

Another study found that mothers desire to be given information about newborn screening prior to birth, yet a majority of them had no recollection of receiving it at any point during or after pregnancy (Hasegawa et al. 301). This population of mothers also largely expressed disapproval of screening for certain conditions that develop after infancy, especially

due to the psychological impact of knowing this information (Hasegawa et al. 302). These results indicate that newborn screening in its current state may not coincide with the views of the people it aims to serve.

Justification for the overall lack in consent gathering involves the necessity of screening to detect conditions that could prove fatal or disabling if not detected and treated soon after birth (Zacharias et al. S39). However, these conditions are typically rare, and most parents encounter a multitude of healthcare professionals throughout pregnancy where screening processes could be explained. Recent challenges to these practices have resulted in states altering their methodology to ensure respect for autonomy and privacy, especially with the expansion of screening programs to include methods like genetic testing (Downie et al.).

To carry out many forms of genetic testing, informed consent must be obtained from the participant. This can be achieved through a variety of processes, and the presentation of the risks and benefits associated with the services offered may influence the participant's ultimate decision. Opt-out approaches employed by many states for mandated screening programs, many of which conduct some genetic testing procedures, have raised questions regarding the legitimacy of the consent obtained from parents (Ross 313).

Some may consider mandatory newborn screening as an overstepping into the private lives of citizens by dictating how parents decide to handle their children's health, especially regarding the expansion in conditions tested over time (van der Burg and Oerlemans 176). Because the refusal to pursue treatment for a possibly disabling or fatal condition may be considered an infliction of harm onto a child, proponents of mandatory screening argue that the wellbeing of a child holds more weight than parental objections (Zacharias et al. S39). This argument has also been used to justify the lack of informed consent associated with newborn

screening in some states, as demonstrated by New Jersey's current practices that have been legally challenged (Superior Court of New Jersey).

For this justification to remain applicable, newborn screening programs would have to test only for specific diseases that are treatable due to early detection (van der Burg and Oerlemans 177). Additional activities performed using samples collected from newborns would also have to be justified by the fact that they would provide immediate benefits to a child's well-being. As demonstrated by recent debates, the current state of newborn screening exceeds these justifiable boundaries and calls for the implementation of thorough informed consent processes.

Approaches to Informed Consent

An opt-out approach to consent has been widely adopted by state newborn screening programs, so samples are expected to be collected from all infants born within the state unless a specific exemption has been requested by parents (Rothwell et al., "The Influence" 231). Debate surrounds the effectiveness of such methods compared to opt-in consent, with scholars citing issues with participants possibly failing to have been presented the materials required to make an informed decision (MacKay 833). In states like New Jersey, where an informed consent process is not in place, parents may not even be aware of newborn screening programs before samples are collected from their children (Superior Court of New Jersey).

Despite current newborn screening programs covering a plethora of conditions, most opt-out approaches only allow for parents to completely withdraw consent instead of selecting for the testing of a subset of disorders (Ross 313). Since some tested conditions are life-threatening and require treatment while others may just provide a diagnosis without a known treatment plan, this method poses problems with the decision-making power of parents. If parents ultimately choose

to opt-out due to their opposition to a subset of tests, the diagnosis of a potentially disabling condition could be missed and have detrimental effects on their child. On the other hand, ultimately opting-in despite opposition to some tests would undermine the parents' autonomy and require exposure to higher-risk testing.

Opt-in methods are seen with processes like registration for organ donation, where people are asked to become a donor, commonly while applying for a driver's license (MacKay 834). These policies require that individuals actively engage with the process before consenting to it, aiming for complete understanding of the material in order to secure true consent. Conversely, opt-out policies may not ensure consent as individuals may bypass the education needed to fully understand the processes they participate in (MacKay 834). In some cases, people may be completely unaware of the possibility to opt-out or of the procedure as a whole.

The use of an opt-out approach may not be problematic in itself, however. A 2016 study determined that pregnant women who received educational materials about the retention of blood spots were more likely to be trusting of newborn screening processes and to support an opt-out approach (Rothwell et al., "The Influence" 233). Participants in genetic research regarding cancer have also indicated that they would prefer to grant re-consent when their data is used in research on another health condition or transferred to another institution (Goodman et al. 224). This suggests that informed consent processes that ensure the understanding of procedures may be absent at multiple points in many state-mandated newborn screening programs.

The use of resources for opt-in approaches is also a source of concern since healthcare procedures are fast-paced, especially during the peri-partum period (Rothwell et al., "The Influence" 235). However, this issue can be mitigated since educational materials do not have to be presented near birth. *Beleno* has illustrated how a lack of transparency surrounding newborn

screening practices can result in the loss of a plethora of valuable resources for advancing public health (Lewis et al. 704). Neglecting to respect parental autonomy poses a larger threat to maintaining long-term public trust in screening programs, which could negatively impact their functions in public health.

Evaluating Risk

Beneficence has to do with the protection of humans from harm while advancing their well-being; this ethical principle emphasizes the importance of balancing risk (The Belmont Report). For example, this principle plays an important role in trials for evaluating medications that could treat debilitating conditions since there are always risks for serious side effects (The Belmont Report). This concept can be expanded to the social impacts of large-scale programs, such as the inappropriate access to samples by outside institutions and the promotion of public health through their operations. As with autonomy, the application of beneficence is largely dependent on the type of procedure being done and the participants.

Screening programs are largely considered low-risk due to the lack of immediate concerns with sample collection and testing, but problems may arise due to tasks performed with stored samples years after their collection. State-mandated programs are justified through the benefit they provide in assessing the health of individuals and its wider impact on advancing public health (Zacharias et al. S39). The expansion of testing has presented risks over time, and these risks may compound with additional developments of genomic methods. The lack of consent processes that distinguish types of testing presents a possibility for harm, as parents may disapprove of a certain set of tests and choose to opt-out of screening as a whole (Ross 314).

Foregoing basic screening could result in neglecting to detect a condition that results in detrimental health effects without treatment.

The risks and benefits associated with a procedure may be viewed differently depending on the individual, further emphasizing the importance of individual decision-making. Researchers may support the use of residual blood spots for secondary research in order to power scientific advancement, but participants may view this as a threat to their privacy (McCulloch 333). A variety of factors are at play when it comes to how the pros and cons of participation are evaluated. Newborn screening is a somewhat unique area of study since future use of samples could impact the family members of the specimen donor without their consent or knowledge (Downie et al.).

The Right to Privacy

Privacy pertains to the ability of individuals to exhibit control over the dispersal of their information (Knoppers and Abdul-Rahman 99). Because personal information often must be shared with outside entities, especially regarding healthcare organizations, legal and ethical safeguards have been implemented. These protections aim to prevent negative impacts on people's lives due to inappropriate access to their data through breaches or third-party sharing. With an increasing emphasis on the genome's use as a tool to further public health research, these traditional approaches to maintaining privacy have been dismantled. Additionally, the special treatment of genetic information due to its familial nature has ignited discussions about the application of privacy to this type of data (Kilbride 10).

Access to residual blood spots by outside entities, such as law enforcement agencies and research institutions, has ignited concerns regarding how privacy is maintained within newborn

screening. Like with informed consent processes, states have established variable levels of accessibility to residual blood spots. In some states, the Department of Health is responsible for deciding which researchers can obtain access to blood spots, while others specify the types of research projects for which samples can be accessed (Lewis et al. 705). Others neglect to establish guidelines for secondary research at all (Lewis et al. 705). Additionally, the use of genomic testing for newborn screening, especially whole genome sequencing, could amplify already-existing concerns regarding the scope of conditions tested and the amount of information accessible by outside entities (Goldenberg and Sharp 461).

Views of Genetic Information

Genetic exceptionalism refers to the belief that genetic information is unique and must be treated differently than other types of medical information (Domaradzki 105). The wide-reaching impact of one variant within one person's genetic code may justify the need for extra or targeted protection of this information. Parents should be fully aware of the processes involved in collecting and disseminating their child's genetic information because the findings generated from tests may reveal facts about their own health (Kilbride 11). This is consistent with the goal of screening programs to improve public health, and transparency could ultimately enhance participation and engagement with the process. At the same time, privacy protections must be upheld to prevent negative impacts on those who provide genetic information.

Such a large emphasis is placed on genetic testing due to the prevalence of medical conditions linked to genetic variations (Domaradzki 105). With advancements in sequencing methods in recent years and the genomics projects that have emerged due to them, public health initiatives have rapidly evolved to involve genetics. Newborn screening is no exception to this

trend, with the expansion of testing for the goal of detecting and treating conditions as early as possible.

The reality is that most conditions develop from an interplay of many genes and one's environment. Advancements in instrumental methods have expanded the scope of newborn screening which in turn provides samples that are used to promote the understanding of the genome (Wolf et al. 7). Hindering any part of this cycle could negatively impact scientific advancements; however, the unmonitored dispersal of sensitive genomic information poses threats to privacy.

The definition of genetic information is a common source of controversy when it comes to addressing ethical issues in the realm of genetic testing. Federal laws have outlined regulations for what certain entities can do with genetic information (Rothstein 5). However, the extent of these protections is not always clear. GINA defines genetic information as the genetic tests of an individual or that individual's family member, which may introduce ambiguity when it comes to more complex sources of genetic information, such as residual blood spots (Rothstein 5). This trend can be detected in many policies pertaining to genetic information, from the federal level to local laws. This ambiguity can result in inconsistencies with the decisions made regarding ethical issues with genetic data, as illustrated by *Bearder*.

Debate surrounding the classification of genetic data in legal and ethical contexts has resulted in uncertainty and concerns surrounding the privacy protections that are provided for individuals who participate in genetic testing. The effectiveness of security practices within genetic testing raises more questions regarding how data is protected beyond guidelines. On the other hand, some argue that people with conditions have the "duty to warn" close relatives about their risk (Sommerville and English 147). Under this lens, genetic information may not be

private at all, and questions arise regarding the scope of what can be done with accessed data. The extent to which genetic information must be disseminated within a family may depend on the condition, introducing complexity to the issue of privacy (Sommerville and English 148) .

De-identification of Samples

Some states have implemented practices such as de-identifying samples before they are utilized in research, but samples have become easier to identify due to advancements in genomic technology (Lewis et al. 708). As a result, family members of newborns may have their privacy at risk since a residual blood spot may have the information needed to track down a relative, as shown by *NJ OPD et al.* (Superior Court of New Jersey).

The Health Insurance Portability and Accountability Act (HIPAA) outlines regulations surrounding the use of protected health information by covered entities (Matranga 459). The concept of de-identified data is a critical factor in these laws, as the removal of identifiers from health data exempts its coverage by HIPAA (Matranga 460). Many states have enacted additional laws prohibiting providers from sharing information regarding conditions that are highly stigmatized, such as HIV/AIDS, substance abuse, and genetic disorders, but they do not fully address de-identified data (Saks et al. 414).

Regarding identifiable biospecimens, consent from the donor is typically required (Schmidt and Callier 304). Many rules that apply to identifiable samples no longer apply when specimens are anonymized, reducing restrictions for access and overall privacy protections due to a belief that the risk for harm is greatly decreased. In reality, the distinction between anonymized and identifiable samples is not always clear. The transfer of data between institutions of different areas of expertise increases the possibility of unintentional or purposeful

re-identification (Schmidt and Callier 305). The de-identification of data originally intended to protect individuals may ultimately pose a larger threat to privacy due to the loosened accessibility regulations associated with this form of information.

Genetic information is inherently familial, meaning that what is revealed by newborn screening also applies to the infant's relatives in some capacity (Kilbride 10). The sharing of data collected by newborn screening presents a challenge beyond the privacy of solely the infant, as a multitude of relatives can be traced using a single sample. The databases to which de-identified data is compared does not need to be genetic; it is possible to determine the identity of genetic information from sources like voter registration or hospital records, though the risk is not as pronounced (Hoffman 1774). The de-identification of residual blood spots does not necessarily provide complete privacy protections for the newborns they were collected from, as genetic sequencing and comparison to databases can permit the re-identification of such data (Hoffman 1764). This demonstrates the need for restricted accessibility when it comes to these samples in order to preserve privacy and reduce the risk of harm.

Security Practices

Security measures taken by state departments are also crucial to maintaining the privacy of newborns and their relatives. Data breaches are common for electronic medical records, with millions of individuals having their information exposed yearly (Arnold 561). With this method of documentation becoming the norm in recent years, privacy risks due to inappropriate access are more prevalent than ever. These issues intertwine with informed consent processes since parents in many states could have their child's privacy compromised without having made a decision to participate in newborn screening.

Millions of electronic health records are compromised annually despite the presence of security mechanisms (Konnoth 1335). The risk of inappropriate access is always present with any type of database, including stored data from newborns. The genetic information originally collected from a newborn could be acquired and used for nefarious activities, such as fraud or insurance discrimination, without the parent even being aware of this risk (Konnoth 1336). This problem is compounded when considering that the majority of parents in the United States do not have a say in their child's data being stored and used for secondary purposes. A large data breach may reveal these previously unknown risks to the public, igniting distrust in biomedical research (Konnoth 1347).

Informed consent procedures would inform parents of these risks and allow them to weigh them along with the benefits of screening. Trends in research show that most people are open to providing consent for the use of their data when asked, but they also do not approve of its nonconsensual use (Goodman et al. 224). People seem to focus most on the maintenance of their autonomy through consent rather than possible risks, so addressing this issue from the front-end could mitigate concerns beyond the acquisition of initial consent. The strengthening of legal protections regarding what entities can access newborn screening data and what actions can be performed on samples could further establish public trust and mitigate risks for harm.

Achieving Justice

The Belmont Report also defines the principle of justice as the equal treatment of individuals through the access to benefits and protection from harms. Regarding this concept, procedures such as medical trials for the treatment of specific disorders should be offered to those with the condition, and respect for autonomy prioritizes the informed decision of the

potential participant (The Belmont Report). Injustices may arise even when the selection process for participants is considered fair. This is because systemic inequities ubiquitous throughout society affect accessibility to such procedures, creating discrepancies among racial groups and other demographics.

With the rise of precision medicine in recent years, disparities have developed between populations regarding access to genetic testing and treatment options for screened conditions (Mamun et al. 517). Mandated newborn screening provides a bridge for closing these gaps by providing services to all infants born within the country. However, this universal approach has largely resulted in the departure from traditional ideals of parental consent and privacy in many states (Ross 309). These concerns are magnified when considering that outside entities may access samples and perform tasks with them unbeknownst to parents.

Newborn screening is perhaps the most accessible form of testing within the country, providing every parent with the opportunity to know about any genetic disorders their child may have and treatment approaches to avoid negative effects later in life. In a way, the universality of newborn screening provides justice for the American population, yet variations in the quality and accessibility of follow-up procedures and treatments for infants with detected conditions may hinder this aspect (Ross 309). Additionally, actions taken with samples beyond testing may disproportionately raise risks for certain populations (Enriquez-Sarano 2347).

Within minority populations, distrust in healthcare is more common and presents a threat to participation in procedures that use an opt-in approach to consent (Rothwell et al., “The Influence” 235). This presents a tension between the ethical principles presented, as a methodology that advances autonomy may exasperate issues with underrepresentation and health-related harms. On the other hand, the current lack of transparency associated with opt-out

policies could result in public distrust if negative impacts of screening receive media attention. Current events in New Jersey have shown that organizations are interested in presenting newborn screening practices to the public, which could result in a resistance to participation and a harmful effect on public health (Superior Court of New Jersey). Thorough education is an important component of opt-in approaches, shown to increase trust in procedures and a willingness to participate (Rothwell et al., "The Influence" 233).

The Need for Transparency

Many of the actions performed using samples from newborns are obscured from the public view due to the framing of the processes used to obtain consent and the overall omission of details regarding the transfer of data among entities. The differences among state regulations for newborn screening programs make it difficult to map out how newborn screening programs function throughout the nation (Lewis et al. 703). The opt-out methodology often used for these initiatives also has resulted in limited public knowledge regarding the use of samples beyond initial testing (Rothwell et al., "Secondary Research" 1474). Achieving true informed consent is not possible without transparency in all processes.

The variability in state policies regarding informed consent and accessibility of samples, along with the uncertainty of privacy risks, may cause distrust in the newborn screening programs that are responsible for the early detection of treatable conditions and the advancement of research (Lewis et al. 708). As a result, parents may choose to opt-out of programs at higher rates with public knowledge of the risks associated with them. This could be detrimental for newborns born with undetected conditions, and the progression of research could be hindered

overall. This demonstrates a need for transparency in all practices associated with newborn screening, which envelops a variety of ethical concerns.

Addressing Informed Consent

The addition and refinement of informed consent processes for newborn screening programs would require decisions to be made about what and how information is presented, as well as what choices are provided to parents (Rothwell et al., "An Assessment" 340). Although the presentation of nuanced information is an important part of informed consent, documents must also be formatted in a way that they are easily understood by the average American adult in a reasonable amount of time. The examination of typical informed consent forms has revealed that a significant amount of extraneous and redundant information is present (Corneli et al. 16). Lengthy informed consent documents may discourage the full comprehension of processes, preventing the acquisition of true consent from participants.

To ensure that consent is effectively obtained from participants, it is necessary that information is presented in a straightforward manner that cannot be easily overlooked. Regarding opt-out approaches, it must be made clear that testing will be carried out without an exemption since parents may assume that screening is opt-in, especially if they lack prior knowledge. Many state-mandated newborn screening programs fail to achieve this, even when information is presented directly to parents.

Opt-in policies may be more likely to obtain actual consent from participants than opt-out policies through the direct engagement with parents through educational materials (MacKay 835). As demonstrated by Minnesota, the implementation of opt-in policies specifically for additional screening and secondary research is possible. This process requires that parents

receive the education necessary to comprehend the scope of newborn screening before making a decision, strengthening respect for autonomy in regard to actions performed with their child's data that diverge from traditional criteria for screening.

Pre-Screening Counseling

The implementation of informed consent practices in all newborn screening programs throughout the country would require the education of participants prior to testing. Genetic counseling is a clinical approach to promoting health literacy for people at-risk of developing a genetic condition or having a child with one (Syurina et al. 202). This methodology has not been applied to newborn screening, perhaps due to the large scope of programs and a lack of resources. Nonetheless, it is important that health literacy is established throughout the general public so that they can understand the best way to approach their own health as well as their children's.

A lack of parental education due to low efforts by professionals to provide the information necessary to understand newborn screening has contributed to confusion and ethical concerns among participants (Hasegawa et al. 301). This opacity regarding the processes of newborn screening has to do with it being mandatory, creating an approach that largely undermines parental knowledge and consent for the purpose of advancing public health.

Widespread initiatives that aim to educate the general public about newborn screening programs could reduce confusion about the process and increase overall trust and willingness to participate. When people are knowledgeable about an important topic and possess decision-making power over it, ethical dilemmas are generally less likely to arise. Counseling prior to testing would help ensure that parents understand how testing occurs and how results could

impact their lives (Syurina et al. 205). Because a variety of conditions are currently screened, some with established treatment plans and others that are not understood well enough to treat, professionals should manage the expectations of parents regarding this range of possibilities (Johnston et al. S16).

One possible way to remedy intrusion by mandated screening programs is to employ a tiered system that requires screening for a strict subset of treatable conditions with early intervention and presents the option of additional voluntary screening (Ross 314). This approach would likely result in an increase of parental awareness regarding newborn screening as a whole while also granting back some decision-making power. The implementation of informed consent for state-mandated screening could also increase public trust in healthcare procedures with a minimal reduction in participation rates.

Pre-Screening Educational Materials

All states require that educational materials are presented during the consent process, with the aim to provide the resources necessary for parents to make an informed decision about participation in screening despite the lack of an opt-in consent form (Schweers 872). However, these materials are often given with a plethora of other information that tends to be overlooked during the stress of the peri-partum period (Botkin et al. 121). Of the states that distribute educational materials about newborn screening, only a small portion of them require that parents be notified of their dispersal (Lewis et al. 707). It has also been found that many of these materials do not provide information regarding the secondary uses of residual blood spots, so parents would not be able to obtain complete knowledge from testing institutions even with thorough educational practices (Lewis et al. 707).

Concerns regarding the lack of informed consent for newborn screening were present within the 1970s, and a 1982 study found that the process of obtaining consent often took less than five minutes and that a very small number of mothers refused testing (Ross 304). This demonstrates that implementing an informed consent process would likely have posed little cost to hospital procedures or newborn screening rates while granting respect to the parents' autonomy and reducing opacity. The scope of testing has expanded since this study was performed, so obtaining consent would likely be a longer process today.

A possible approach to avoid the hindrance of peri-partum procedures is to incorporate education and consent into the appointments leading up to birth (Rothwell et al., "The Influence" 235). To ensure that parents maintain consent at birth, a healthcare professional could confirm the choice shortly before samples are collected; this likely would not take a significant amount of time with education being provided prior to this point. In addition to education provided before birth, post-natal discussions of newborn screening with parents could be implemented to ensure understanding and confirm consent (Rothwell et al., "The Influence" 235). A recent study has demonstrated that parents are less likely to refuse newborn screening when they possess more knowledge about the process, so the education of parents before obtaining consent would likely not have a significant impact on the proportion of infants tested today (Rothwell et al., "The Influence" 234).

Participation by parents in newborn screening is often minimal, resulting in a deficit in knowledge and trust of such processes (Hiller et al. 1284). With the widespread absence of informed consent processes within newborn screening, further ethical concerns emerge for genetic testing specifically. A lack of transparency from healthcare professionals regarding the probabilistic nature of such tests as well as the risk of procedures could entice distrust in the

general public. The continued storage of residual blood spots without consent in many states will also likely result in further lawsuits and the destruction of valuable databases for the promotion of public health. The addition of informed consent processes that ensure parental education would likely not cause a significant change in newborn screening participation while also giving parents the power to make decisions for their children.

Minnesota is a state that requires healthcare professionals who conduct screening to provide parents with a document and notify them about the process before it occurs (2022 Minnesota Statutes). The state has also implemented another layer to the consent process, requiring that parents opt-in to additional research performed using blood samples (2022 Minnesota Statutes). Additionally, the ability of parents to revoke consent for the use and storage of residual blood spots is an important aspect of the process, as the evolution of research methods and information sharing could impact lives years after consent was given under initial conditions (Lewis et al. 706). Addressing the concept of consent at multiple steps in the process ensures that parents are fully informed when making decisions and retain their autonomy for the entire time their child's genetic information is in the possession of the state.

Privacy Policies

Variation exists across states regarding what entities can obtain access to newborn residual blood spots under certain circumstances. Legal cases have arisen surrounding the use of these spots for reasons beyond public health, such as forensics, due to possible inappropriate access. Many states have not enacted laws pertaining to the storage of residual blood spots, and only a small portion require parents to be given information regarding their storage (Lewis et al. 704). A vast majority of states allow for residual blood spots to be used for secondary research,

yet only a small portion of them require that consent be obtained from parents before their use (Lewis et al. 704). For example, a newborn screening program in Texas was sued in 2009 for handing over 800 residual blood spots to the US Armed Forces Pathology Laboratory for forensic purposes without parental consent (Lewis et al. 704). Concerns have only grown larger since, with states altering their newborn screening programs and current lawsuits underway.

To permit the sharing of data between research institutions without compromising the privacy of individuals, protections should be applied at multiple steps in the process. On the federal level, HIPAA does not provide sufficient protections due to the exemption of coverage for de-identified data (Hoffman 1764). In addition to this exception, HIPAA does not prohibit or even discourage attempts to re-identify data (Hoffman 1792). Permitting the access of institutions only if they meet predetermined guidelines could mitigate risks for inappropriate access of genetic information.

The ADA was enacted before the evolution of genetic testing into what it is known as today. This law prevents discrimination based on mental or physical disability, yet it does not account for differential treatment based on predispositions to these conditions (Horvitz and Mulligan 253). Testing and research involving genetic information continue to evolve at a rapid pace, while protections are provided by laws enacted several years or even decades ago. This grants the opportunity for entities to find gaps in these protections and take advantage of individuals' genetic information.

Biobanks utilize access committees to determine which entities may gain access to the database for research purposes (Fortin et al. 106). As seen with many aspects of data sharing associated with genetic information, these committees are integral to establishing a balance between the promotion of research and the protection of stored data (Fortin et al. 108). Some

biobanks will require that the institutions that receive access to databases return results in order to monitor the actions being taken with data (Fortin et al. 109). Access to residual blood spots could perhaps be regulated in a similar manner, ensuring that all secondary uses of transferred data are appropriate and within the scope of public health.

Conclusion

Approaches to consent and privacy protections among state newborn screening programs are highly variable, producing inconsistent adhesion to ethical principles that aim to provide sufficient protection for parents and their newborns. Many states have adopted opt-out policies that presume consent, but this is not necessarily applicable to newborn screening programs since parents are often not knowledgeable about the process, especially beyond initial sample collection and testing (MacKay 834). The use of residual blood spots for secondary purposes by most states has ignited privacy concerns, especially regarding research by outside entities and access by law enforcement agencies, as demonstrated by *Bearder* and *NJ OPD et al*.

Legal frameworks for newborn screening programs are highly fragmented and inconsistent on a federal and state level. Concerns about possible gaps among laws allowing for the violation of privacy by certain entities like law enforcement agencies have arisen, indicating the need for strengthened protections. The incorporation of informed consent processes within state newborn screening programs, such as Minnesota's current approach, would likely eliminate much of the mystery surrounding the process in most states. This approach would involve the education of parents regarding newborn screening itself as well as the actions performed with stored samples, effectively reducing opacity. Mothers have previously expressed openness to opt-out approaches to consent after receiving thorough education about newborn screening

procedures and residual blood spot uses, although opt-in approaches that allow nuanced consent may provide more respect for parents' autonomy (Rothwell et al., "The Influence" 233).

Privacy concerns would likely be greatly reduced for parents through the use of informed consent, but inappropriate access to residual blood spots remains an issue, as shown by *NJ OPD et al*. Restricting access to blood spots to predetermined areas of research could work against possible breaches of privacy, reducing the risk for harm of newborns and their relatives (Hoffman 1774). Laws such as HIPAA and GINA have also not been updated to match the rapidly evolving area of genetic testing, so protections could be strengthened on a federal level as well (Matranga 460).

Prioritizing the education of the public in order to build health literacy and encourage informed decision-making is necessary for upholding autonomy within newborn screening (Newcomb et al. 293). Informing parents of the benefits, risks, and full scope of newborn screening is essential to reducing opacity and encouraging trust in these crucial public health initiatives. Organizations like the New Jersey Monitor, a news site and plaintiff in the *NJ OPD et al.* case, are interested in revealing practices to the public that present ethical challenges within newborn screening (Supreme Court of New Jersey). This may be the push needed to call for policies that strengthen consent processes and privacy protections across the nation. It is imperative that the current state of newborn screening in many states is altered to respect parental autonomy and the right to privacy; delaying this process could be detrimental due to the risks of further breaches in privacy and increasing distrust in public health research.

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