The Down Syndrome Information Act: Balancing the Advances of Prenatal Testing Through Public Policy

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Abstract
Since the dawn of prenatal testing in the 1970s, concerns have been raised over its administration to respect a mother's autonomy as well as the expressive critique against those with the tested-for condition. Advances in prenatal testing have made it such that more mothers than ever are given a test result of Down syndrome, yet are not provided the rest of the information recommended by professional guidelines. In response, first federal legislation and then, increasingly, state legislation is requiring that this information be provided to expectant mothers. Though receiving broad bipartisan support in passage, some of the statutes have received criticism. These public policy measures will be surveyed and evaluated as to their relative merits and limitations.

Key Words: Down syndrome; prenatal testing; legislation; ethics

In the past decade, the field of prenatal genetic testing has experienced dramatic advances with Down syndrome testing being at the forefront. As these technologies have evolved, so have professional medical recommendations on their use. At the same time, and over an even longer period of time, advances have been made in the lives of those living with Down syndrome. However, the challenge of keeping medical providers and patients up-to-date on both has been persistent since the dawn of prenatal testing in the 1970s. To address this challenge, federal legislation was enacted in 2008 and beginning in 2012, state initiatives have resulted in over a quarter of the states enacting legislation of their own. Though receiving broad bipartisan support in passage, some of the statutes have received criticism. These public policy measures will be surveyed and compared for similarities and distinctions in legislative language, followed by a discussion on the need for the legislation, the criticism some of the statutes have received, and the relative implementation for the various state measures.

Methods
The federal and state legislation are a matter of public record and each was reviewed for content with a follow-on survey of each measures implementation by the responsible state agency. The author has been involved in the passage of almost all of the initiatives discussed. He was involved in the grass roots advocacy for the passage of the federal legislation. He led the effort for passage of the state version of the legislation in his home state of Kentucky. He has co-presented with the executive director of the Massachusetts Down Syndrome Congress (MDSC) and representatives of the National Down Syndrome Society's (NDSS) policy center at an annual conference since 2013 to train local Down syndrome organization leaders on how to pass the legislation. Monthly, he serves as a moderator for a national call where, among the matters discussed by participants, are the status of efforts in their respective states. This background and involvement further informs the results and discussion herein.

Results
The “Kennedy-Brownback” Act
In 2005, the Prenatally & Postnatally Diagnosed Conditions Awareness Act was introduced in its first iteration as a bill. Madeleine Will, then director of NDSS' policy center, and a mother whose son was born with Down syndrome, spearheaded the effort. Any discussion of prenatal genetic testing inevitability will involve a discussion of abortion. Sen. Ted Kennedy, an ardent pro-choice advocate, and Sen. Sam Brownback, an
equally strident pro-life advocate, were the lead co-sponsors to demonstrate that the purpose of this law was not to expand reproductive rights nor limit abortion. Instead, the purpose of the law was to ensure expectant and new parents received accurate, up-to-date information on the nature of the testing that was conducted and on the conditions that were detected.

Hearings were held in which the research of Dr. Brian Skotko was featured. Dr. Skotko, whose sister has Down syndrome, had conducted surveys of mothers who had received the diagnosis that their child had Down syndrome (Skotko 2005a, 2005b). The overwhelming conclusion was that the way the diagnosis was delivered needed improvement in both the method of delivery and the accuracy of the description of a life with Down syndrome.

The initial version of what became known as the Kennedy-Brownback Act required the delivery of up-to-date, accurate information about the nature of the testing and the condition diagnosed with a funding provision of $5 million for each of 5 years. Being federal legislation, it took several years to work its way through the legislative process. Two years after being introduced, prenatal testing underwent a historic change.

In 2007, the American College of Obstetricians and Gynecologists (ACOG) changed its recommendations. Previously, only women 35 or older had been recommended to be offered prenatal testing for aneuploidies, conditions involving an extra or missing chromosome, the most common of which being Down syndrome. ACOG changed its recommendations such that all women were to be offered both prenatal screening and diagnostic testing (ACOG, 2007). This added to the urgency of passage of the Kennedy-Brownback Act, because the number of women to be offered prenatal testing expanded from 20%—the percentage at that time of pregnancies carried by women 35 or older—to 100%.

Compromises were made in order to secure a sufficient number of votes for the bill’s passage into law. These compromises stripped the mandatory language from the bill and instead merely authorized the Secretary of Health & Human Services (HHS) to provide grants for the development of patient education resources (Pub. L. No. 110-374, 2008). The bill was passed by unanimous voice vote in both the House of Representatives and the Senate. Since its passage, HHS awarded a grant of approximately $400,000 for 2 years to Genetic Alliance. The grant then was cut in half to just $200,000 for 1 year and nothing of note came of it. No further grants have been made.

State-Level Initiatives
Virginia passed an informed consent law in 1995 requiring women to receive adequate information to make informed choices about genetic testing (Va. Code Ann. Sec. 54.1-2403.01). In 2008, prior to the passage of the Kennedy-Brownback Act, Missouri passed a bill introduced by state Senator John Loudon, a father to an adopted son with Down syndrome, which required the delivery of information to patients and provided funding for its implementation (Mo.Rev.Stat. Sec. 191.923, 2007).

Since the passage of Kennedy-Brownback, 12 states have passed laws having various titles but what will be referred to here as the “Down Syndrome Information Act” or “DSIA.” In 2012, Massachusetts led by passing the first DSIA (Chapter 126 of the Acts of 2012), with Florida soon following afterward that same year (Sec. 383.141, Fla. Stat. 2012). Massachusetts was prompted by the development of the latest prenatal genetic testing technology: cell-free DNA testing, which entered the commercial market in October 2011. Initially, ACOG issued a position statement recognizing cell free DNA as a viable screening test with the limitation of it being offered only to mothers considered high-risk (ACOG, 2012). ACOG removed this restriction in the summer of 2015 such that all women may be offered cell free DNA testing, adding to the urgency of DSIA’s passage (ACOG, 2015).

In 2013, Kentucky joined Massachusetts (Ky. Rev. Stat. 211.192). In 2014, a geometric rise in the number of states occurred with Delaware, Maryland, Louisiana, Pennsylvania, and Ohio all passing versions of the DSIA (House Bill 1058; Maryland Senate Bill 654; La. Rev. Stat. Ann. 40:1300.392; Pub. L. 2450, No. 130, Cl. 35; Ohio Rev. Code 3701.69). In 2015, Texas, Indiana, Minnesota, and Illinois all passed versions of the DSIA (H.B. No. 3374; IC 16-35-9.2; Minn. Stat. 145.471; HB 3158). These various DSIA’s will be discussed grouped in categories by the similarities they share.

Model, Mandatory Language
Massachusetts, being the first, has served as the model upon which all others have based their DSIA’s language. Therefore, the full text of what’s
required by the Massachusetts DSIA will be quoted in full:

(a) For the purposes of this section, the term “Down syndrome” shall mean a chromosomal condition caused by an error in cell division that results in the presence of an extra whole or partial copy of chromosome 21. Any facility, as defined in section 70E, physician, health care provider, nurse midwife or genetic counselor who renders prenatal care, postnatal care or genetic counseling, upon receipt of a positive test result from a test for Down syndrome, shall provide the expectant or new parent with information provided by the department under subsection

(b) The department shall make available to any person who renders prenatal care, postnatal care or genetic counseling of parents who receive a prenatal or postnatal diagnosis of Down syndrome the following:

(1) up-to-date, evidence-based, written information about Down syndrome that has been reviewed by medical experts and national Down syndrome organizations. The written information provided shall include physical, developmental, educational, and psychosocial outcomes, life expectancy, clinical course, and intellectual and functional development and treatment options; and

(2) contact information regarding First Call programs and support services, including information hotlines specific to Down syndrome, resource centers or clearinghouses, national and local Down syndrome organizations such as the Massachusetts Down Syndrome Congress, and other education and support programs. The department may also make such information available to any other person who has received a positive test result from a test for Down syndrome.

(c) Information provided under this section shall be culturally and linguistically appropriate for women receiving a positive prenatal diagnosis or for the family of a child receiving a postnatal diagnosis of Down syndrome.

The key provisions of the DSIA are:

[1] Broad definition of medical professionals who are to provide the information;

[2] That information is to be provided with a prenatal or postnatal test indicative of Down syndrome, but not limited to necessarily a diagnosis as screening technologies have advanced;

[3] That the information is up-to-date, reviewed by medical professionals and Down syndrome organizations, and covers a spectrum of information, not just medical information;

[4] Contact information will be provided to local Down syndrome support organizations; and,

[5] The information is to be culturally and linguistically appropriate for patients.

All of the states retain these basic elements in each of their respective DSIsAs. For instance, Kentucky's DSIA is taken almost verbatim from Massachusetts, with the exception that local Down syndrome support organizations in Kentucky are substituted for MDSC's First Call program. Pennsylvania makes it explicit that all that is required of the state agency is for the recommended materials to be made available online. Ohio states that patients should receive a “sheet” containing the recommended information and support resources. Florida, Indiana, and Minnesota do not limit their acts to just Down syndrome, but cover all conditions; Kentucky's DSIA was amended in 2015 to add spina bifida. Florida further empanels a council to make recommendations on the information to be provided about the variety of conditions that may be prenatally or postnatally detected. But, content-wise, Massachusetts remains the model for the general elements of the DSIA. A minority of the states with DSIsAs, however, have made key modifications.

Permissive, Not Mandatory, Language
Two of the states with DSIsAs made a key compromise in the Act's requirements. Maryland and Illinois both removed the mandatory language requiring that health care professionals provide the recommended information with the delivery of a test result. Instead, both require the responsible state agency to identify the recommended information and referral resources and make them available to practitioners and
patients, but that is all that is required. In Maryland, health care professionals then are simply “authorized” to provide the recommended materials to their patients, but not required. In Illinois, health care professionals “may” provide the recommended information to their patients. Equally, in both states, the health care professional may not choose to provide the recommended information to their patients.

Termination Not Recognized as an Option
Beginning in Louisiana, and then in Texas and Indiana, those states added a key provision to the DSIA:

All information provided pursuant to the provisions of this Section shall be culturally and linguistically appropriate for the recipient of the information, and shall not engage in discrimination based on disability or genetic variation by explicitly or implicitly presenting pregnancy termination as a neutral or acceptable option when a prenatal test indicates a probability or diagnosis that the unborn child has Down syndrome or any other health condition.

In Louisiana, the DSIA was not initiated by Down syndrome advocates but by the Bioethics Defense Fund, a pro-life organization (Bioethics Defense Fund, 2014). In Indiana, Indiana Right to Life (IRTL) was involved in the drafting of the legislation (IRTL, 2015). And in Texas, competing versions of the DSIA were introduced in the same session, with the clause originating from a pro-life representative with the backing of pro-life organizations (Texas Alliance for Life, 2015).

Discussion
Need for Legislation
The premise of the DSIA is to require medical professionals to counsel their patients according to professional medical guidelines. ACOG’s 2007 practice guidelines recommended that patients receive the “natural history” about the tested-for condition and recognized that parent support organizations may be very helpful. Other professional societies have been even more explicit, with the National Society of Genetic Counselors (NSGC) and the American College of Medical Genetics & Genomics recognizing specific patient resources (Gregg et al., 2013; Sheets et al., 2011).

More than one legislator, when briefed on the legislation, has then wondered that if the guidelines already recommend this information, why a law is needed. The answer given is that a law is needed because medical professionals too often do not provide their patients with the recommended information and referral to support organizations.

The year before ACOG’s recommendations, a study found that 45% of obstetricians admitted their training on prenatal genetic testing was “barely adequate” or “nonexistent.” (Cleary-Goldman et al., 2006). Two years after the ACOG recommendations, a survey of 500 ACOG fellows determined that although almost all obstetricians were offering prenatal genetic testing to all patients, in compliance with the guidelines, only 29% stated that they provided educational materials to their patients (Driscoll, Morgan, & Schulkin, 2009). Given this, it is not surprising then that a survey of studies on informed consent for prenatal testing found that the overwhelming conclusion was that the administration of prenatal testing did not respect a woman’s autonomy (Seavilleklein, 2009). As recently as 2013, a survey of mothers receiving a pre- or post-natal Down syndrome diagnosis rated their experience as negative 2.5 times for every one positive experience (Nelson-Goff et al., 2013).

Testimony given in support of the various state measures further supported the need for the legislation and gave voice to mothers’ experiences receiving a Down syndrome diagnosis.

In Maryland, Heather Sachs testified about receiving a postnatal diagnosis in 2006 for her daughter. The information the hospital provided her was a pamphlet entitled, “So You’ve Had a Mongoloid: Now What?” (Ryan, 2014).

In Minnesota, Sandi Holmgren shared what her medical team told her when her daughter with Down syndrome, Mikayla, was born:

Because she was a preemie, we were told that she may never take a bottle and may have to be on feeding tubes for a very long time. She was nursing before she left the NICU.

We were told that she probably would not walk until she was 3 or 4. She walked at 13 months.

We were told she may not start talking until 2 years or later. Her first word was Banana at the
Mikayla Holmgren then testified:

I am 20 years old. I attend St. Croix Preparatory Academy and I am currently in their transitions program. I am a TA in lower school. I love to golf and do gymnastics. I love to paint. (Holmgren, 2015)

The need for legislation requiring that health care providers provide accurate information with referral to support groups was encapsulated in an exchange during a hearing on the Texas DSIA.

Carrie Kauffman, an obstetrician/gynecologist from Austin, Texas, testified representing, the Texas Medical Association, Texas Association of OBGYN, Texas Pediatric Society, Texas Academy of Family Practitioners, and the Texas Society of Genetic Counselors:

Kauffman: Thank you for having me here to speak today. What I would like to address to the committee here is that, we as a broad group of medical professionals working here in Austin, Texas and across the state, oppose this bill due to the mandate that’s included in the language. We support the intent of the bill. We support dissemination of information, education and accurate information. We support supporting our patients, um, however we do oppose a bill that mandates information to be handed to a patient at a specific time-point during their care with us. So I’d like to just stop there and see if I can answer any questions for you guys.

Rep. Farney: Sounds like you support everything about it, except doing it. (Kauffman Testimony, 2015)

Hence, the need for a legal mandate to ensure that women are provided the information professional guidelines recommend accompany prenatal genetic testing.

Criticism

The DSIA has passed unanimously or with near-unanimity in every state where it has been called for a vote. It has been enacted in states with Democrat control of the legislative and the executive branches (e.g., Massachusetts, Delaware), in Republican-controlled state governments (e.g., Texas, Ohio), and in states with divided government between the two major parties (e.g., Kentucky, Illinois). Despite this broad agreement, it has not been without its critics.

Interference With Physician-Patient Relationship

The Pennsylvania law did not draw much criticism while winding its way through the legislature. But since its enactment, it has been criticized for interfering with medical professionals counseling of their patients.

Prompted by the Pennsylvania DSIA, the pro-choice website RH Reality Check criticized the law for requiring that physicians follow a pro-life “script” when counseling their patients (Murtha, 2014). Bioethicist Art Caplan, citing the Pennsylvania version, has criticized the DSIA for “over-turn[ing] the long-standing foundational ethical norm of genetic testing and counseling—neutrality in the provision of information” because Caplan claims that under the DSIA “only positive information is mandated” (Caplan, 2015, p. 3). A simple review of the actual legislation dispels this criticism.

The Pennsylvania DSIA tracks the same language as Massachusetts’ initiating DSIA. No DSIA that has been passed mandates a script that healthcare professionals are required to recite when delivering a test result for Down syndrome. Similarly, no DSIA prescribes that patients receive “only positive information.” The DSIA instead requires professionals to provide the type of information recommended by their professional guidelines, with the exception of the states that are the subject of the next area of criticism.

A Pro-Life Trojan Horse

The Louisiana, Texas, and Indiana’s DSIA have drawn criticism for their provision prohibiting the recognition of termination as an option following a prenatal test result. David Perry, a professor of history and father whose son has Down syndrome, criticized the Louisiana law for driving a “wedge” between disability rights advocates who are pro-choice and those who are pro-life (Perry, 2015).

Distinguishing the typical criticism of pro-life laws that mandate “the inclusion of untrue information,” Perry accused the Louisiana law of demand-
ing “that health-care providers lie by omission instead” by leaving out the mention of abortion as an option.

Under current professional guidelines health care providers are instructed that they “should” counsel a patient about the option of abortion following a diagnosis (ACOG, 2007). In states that recognize the personal injury cause of action referred to as “wrongful birth,” professionals who fail to advise their patients about selective abortion expose themselves to liability of potential multi-million dollar judgments. The pro-life versions of the DSIA place physicians in those states in a dilemma by prohibiting them from providing materials that cover all options.

Nancy Iannone, an instructor at Rutgers Law School whose daughter has Down syndrome, is a co-author of one of the resources recommended by the NSGC. Iannone wondered if measures like that of Louisiana and Indiana would have the exact opposite effect of their intent to prevent women from being advised about abortion, given their physicians’ professional obligation to do so:

[Will providers look for strongly-worded pro-termination information to balance the perceived pro-life information provided by the state? Or will providers talk of termination with whatever bias they already have, be it a pro-life, neutral, or pro-termination stance? (Iannone, 2014)

The addition of the clauses in Louisiana and Indiana’s version of the DSIA have drawn further criticism. As these measures are the result of efforts primarily directed by pro-life organizations—and not Down syndrome or disability organizations—it risks the DSIA being tainted as a Trojan Horse for pro-life legislation (Leach, 2014a). From the inception of the Kennedy-Brownback Act, the DSIA is supposed to be solely a pro-information piece of legislation. The insertion of a clause prohibiting the state from recognizing materials that cover all reproductive options risks the DSIA being seen as a pro-life vehicle misrepresented as a pro-information measure. The insertion of the clause could justifiably critics grouping the DSIA with other anti-abortion measures that mandate patients receive certain information, which are criticized by health care professionals for intruding on the physician-patient relationship (Sawicki, 2011). Should it be so tainted, then the heretofore broad bipartisan support for a pro-information law will degenerate into the logjam of abortion politics, with fewer acts being passed and the status quo of parents not receiving accurate information being perpetuated.

Implementation

All of the DSIA are in their infancy, with most being barely over a year old. Still, trends are already suggesting what works and what does not with the DSIA.

The main factor that is directly related to greater implementation is funding. One of the resources recognized by the ACMG and NSGC and by states that have passed the DSIA is the booklet Understanding a Down Syndrome Diagnosis published by Lettercase and distributed by the National Center for Prenatal & Postnatal Down Syndrome Resources at the University of Kentucky (Meredith, 2013). Research was presented at the 2014 ACMG annual conference showing a direct link between public funds and more resources being provided to practicing obstetricians (Meredith & Sheets, 2014). Similarly, in those states that have not committed public funds, where other funding has been procured to implement the act, more materials have been provided. In Kentucky, no appropriations have been made, but a local charitable foundation awarded a grant for the distribution of the recommended materials to every practicing obstetrician in the state. (Human Development Institute, 2015).

In almost every state that has passed the DSIA, the responsible agency, usually the Department for Public Health, has created a web page with a listing of recommended resources and links to those available on line. This implementation has been hastened when the recommended resources are those already recognized by professional guidelines. For instance, the web sites for Massachusetts and Kentucky were established within mere months of passage of their respective DSIA and track the materials recommended by ACOG, ACMG, and NSGC. On the other hand, in those states that have chosen to create their own materials, the development time for doing so necessarily has resulted in a lag in both implementation and quality. Contra Caplan’s (2015) criticism of the DSIA, the implementation of the Pennsylvania DSIA has not focused on “only positive information.” Instead, what was developed memorialized...
much of what had been the criticism of parents when they received a diagnosis: emphasis on possible associated medical conditions with few pictures or life stories of what a life with Down syndrome can be like (Leach, 2014b). Prompted in part by the subpar information developed by Pennsylvania and due to future states likely passing their own DSIA, the NSGC published a fact sheet to be used as a model for states implementing the DSIA (NSGC, 2015). The fact sheet tracks the NSGC’s practice guidelines which were based on surveys of medical professionals and parents of children with Down syndrome on what information both groups agreed parents should receive with a diagnosis. Finally, in states that have diverged from the professional guidelines, the laws have yet to be implemented. In Louisiana, though the DSIA was enacted in the summer of 2014, nothing since has been done to implement the Act.

No study has yet been done to measure the compliance of any state’s DSIA with no current DSIA requiring reporting to track compliance by professionals. All that does exist is anecdotal information left on web sites. Comments from mothers receiving a prenatal test result for Down syndrome in states with DSIA’s have noted that they were not provided any of the recommended information by the responsible state agency. For example, in 2015, one mother shared

"Additionally, I’m located in Florida, and I read your post on the Down Syndrome Information Act. I see that 383.141 calls for counseling and information to be paired with the delivery of positive results. We did not receive this from my obstetrician’s office. (Leach, 2015)"

As has been shown by many laws, simple passage does not mean they will be followed.

It is expected that so far most medical professionals honor the DSIA in the breach rather than in compliance, and is likely due to simple ignorance of the law’s requirements through no fault of their own. Although press releases and articles are written at the time of the DSIA’s passage, it cannot be expected for health care professionals to change their practice without more awareness provided to them about their respective state’s requirements (see e.g., North, 2014; Ryan, 2014).

Unfortunately, like much of the administration of prenatal testing, it may take a legal threat to increase compliance. The first conventional prenatal screen, the alpha fetoprotein or “AFP” screen, initially was not recognized for detecting Down syndrome. After a wrongful birth lawsuit was brought by a patient who gave birth to a child with Down syndrome after not being offered the AFP screen, ACOG issued its first ever “liability alert” instructing obstetricians to both offer their patients AFP screening and document the offer and the patient’s decision (Suter, 2002). Although damages are unlikely to be recognized by courts, as great as they are for wrongful birth lawsuits, noncompliance with state legislation can give rise to civil claims, complaints with the board of medical licensure, and complaints with hospital patient ombudsmans. And, if in Texas, the last two are the only options because although the Texas DSIA mandates the provision of the recommended information, it bars any civil or criminal liability against practitioners who fail to follow the DSIA’s requirements.

**Conclusion**

Since the inception of prenatal testing for Down syndrome, concerns have been raised over whether it is ethically administered to respect a woman’s autonomy and that it discriminates against those with the tested-for condition. Studies have reported negative experiences of mothers with how their medical professional delivered the diagnosis of Down syndrome. At the same time, prenatal testing has continued to evolve to allow earlier, more accurate assessments of a mother’s likelihood for having a child with Down syndrome. In the face of these developments and the persistent challenge of having medical professionals fully follow the professional guidelines concerning prenatal testing, Down syndrome advocates have been introducing state measures called the Down Syndrome Information Act (DSIA). With each passing year since 2012, more and more states are enacting their versions of the DSIA with various levels of implementation. Future DSIAs can be expected given the broad bipartisan support and near unanimous passage the law has received no matter the politics of the given state. Going forward, implementation will increase, and more mothers can be assured of receiving the full information recommended to accompany a prenatal test result for Down syndrome, should states provide funding and the responsible agencies recognize the resources rec-
ommended by professional guidelines. Keeping abortion politics out of the DSIA will ensure it stays true to its inception as not a pro-life or a pro-choice policy measure, but a pro-information law. Expectant mothers accepting prenatal testing are seeking information. The DSIA’s intent is to ensure they receive the recommended information about Down syndrome and available support resources.

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Conflict of Interest: Materials from the National Center for Prenatal & Postnatal Down Syndrome Resources are discussed herein where I serve as the bioethics specialist.

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