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**Improving Joyful Lives: Society's  
Response to Difference and  
Disability**

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Mark Bradford

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**Abstract:**

Down syndrome is a highly complex genetic condition, but it is also a highly complex sociological phenomenon that is iconic in its significance to questions regarding quality of life, selective abortion, and the extent and legitimacy of research into medical treatment to enhance cognition in the intellectually disabled. As technologies of prenatal screening and diagnosis become more sophisticated, Down syndrome provides an initial test of how society will respond to difference and disability. It has been shown that peer support, as well as accurate and balanced information that exposes the positive experiences of those with Down syndrome and their families, can influence a woman's choice regarding whether to continue her pregnancy. Therefore, it is critical that federal and state laws be expanded to require that this information be provided upon receiving a prenatal diagnosis of Down syndrome. Also, advocates should work to expand prenatal nondiscrimination legislation that is consistent with other federal laws intended to protect the disabled. While research to improve the lives of those living with Down syndrome has progressed rapidly, federal funding for Down syndrome research lags considerably behind other similar genetic disorders. Research to improve birth outcomes and quality of life over the lifespan will certainly improve the message given with a prenatal diagnosis and discourage the termination of Down syndrome pregnancies.

**Introduction**

In the United States, approximately 6,000 individuals are born each year with Down syndrome (trisomy 21). The live birth incidence is about one in 700.<sup>i</sup> Down syndrome is the most common genetic cause of intellectual disability, with the U.S. population estimated to be between 250,000 and 400,000 people, and the worldwide population at six million.

Down syndrome is a highly complex genetic medical condition; it is also a highly complex sociological phenomenon that is iconic in its significance to questions regarding quality of life, selective abortion, and the extent and legitimacy of research into medical treatment to enhance cognition in the intellectually disabled. Iconic, because Down syndrome provides the initial test of how society responds to difference and disability when offered increasingly sophisticated means of prenatal screening technologies in a pro-abortion culture.

The most recent study of abortion following a confirmed prenatal diagnosis of Down syndrome showed that, depending on several factors such as time of prenatal diagnosis, geographical region, ethnicity and religious belief, abortion rates range

from 61% to 93% in the United States.<sup>ii</sup> In France, where prenatal screening has been enshrined in public policy, the rate increases to at least 96%.<sup>iii</sup>

With such a high termination rate, research done by Dr. Brian Skotko, co-director of the Down Syndrome Program at Massachusetts General Hospital, is particularly striking. He has shown that:

- 99% of people with Down syndrome are happy with their lives
- 97% of people with Down syndrome like who they are
- 99% of parents said they love their child with Down syndrome
- 5% of parents felt embarrassed by their child
- 97% of brothers/sisters, ages 9-11, said they love their sibling<sup>iv</sup>

Individuals and families living with Down syndrome overwhelmingly report satisfaction with their lives, but the majority of parents continue to elect abortion following prenatal diagnosis. The contrast presented in these two sources raises critical questions about how prenatal diagnosis is delivered, the perception of support for the intellectually disabled and their families, and the stigma that still remains regarding intellectual disability.

This paper will present a short history of the disability and suggest possible reasons for this stark dichotomy. It will also offer suggestions for how policy makers and advocates might work to create a future where true acceptance and inclusion may be realized for all those conceived with Down syndrome – not just the current minority who are allowed to live.

## **Modern History of Down Syndrome**

### Cause and Discovery

Most typically, Down syndrome is caused by an error in meiotic cell division (usually the mother's oocyte), called a "nondisjunction," in which the 21<sup>st</sup> chromosome fails to divide. At conception when the parents' gametes fuse to create the new unique individual, this individual has three chromosomes 21 rather than the typical two, or a total of 47 human chromosomes in every cell of the body. The presence of an extra chromosome 21, with its full complement of genetic activity, creates a severe metabolic disturbance resulting in mild to moderate intellectual disability, distinctive physical traits, and often more or less serious associated medical conditions, e.g., cardiac defects, leukemia, gastrointestinal issues, various autoimmune disorders, and several others.<sup>v</sup>

In 2% - 4% of conceptions, not all cells are affected and the trisomy is restricted to certain cell lines creating a mosaic effect.<sup>vi</sup> “Mosaic” Down syndrome is typically less severe in its effect than a full trisomy. A third type of Down syndrome, “translocation,” results when part of the 21<sup>st</sup> chromosome breaks off and attaches to a different chromosome. The usual complement of 46 chromosomes remains in these individuals, but because a fragment of chromosome 21 becomes attached to another chromosome, the individual may express some characteristics of the Down syndrome phenotype.

A genetic description of Down syndrome is a fairly recent development. It was first developed in 1958 by Dr. Jérôme Lejeune, a medical doctor and researcher working in Paris, and subsequently published in 1959 by the team of Jérôme Lejeune, Marthe Gautier, and Raymond Turpin<sup>vii</sup>. The identification of the genetic cause of Down syndrome was made possible with the introduction of a new technique that, for the first time, allowed the observation of individual chromosomes under a microscope. Lejeune, using slide preparations prepared by his associate Gautier, was able to enlarge a photograph of an individual’s chromosomes, called a *karyotype*, and cut and paste them together by size to identify the supernumerary chromosome on the 21<sup>st</sup> pair.

Lejeune’s discovery was revolutionary in its importance for the future of modern medicine. Until then it had been commonly believed that Down syndrome was due to some fault of the parent. Many believed that it was caused by a venereal infection and even that it was contagious. These children - children because few lived to adulthood - were kept hidden or placed in institutions because they were a source of shame for their families.

John Langdon Down was the first to apply the term “mongoloid” to those exhibiting the common physical and intellectual features of trisomy 21 in his work, *Observations on an Ethnic Classification of Idiots*, published in 1866. To quote his text:

The number of idiots who arrange themselves around the Mongolian type is so great, and they present such a close resemblance to one another in mental power, that I shall describe an idiot member of this racial division, selected from the large number that have fallen under my observation. <sup>viii</sup>

Down’s term “mongoloid” originated in the medical vocabulary as a descriptive term, along with other terms for the intellectually disabled such as “idiots,” “imbeciles,” and “dullards,” but crossed over into common parlance where it gained pejorative implications in the 20<sup>th</sup> century. Due to its inaccuracy and racial overtones, the term “mongoloid” fell into disrepute in the medical community by the

mid-20<sup>th</sup> century. On April 8, 1961, two years after the publication of the paper describing the genetic origin of “mongolism” by the Lejeune team, a group of 19 prominent scientists, including Lejeune, submitted a letter to the *Lancet* urging the scientific community to abandon the term. Of five suggested alternatives, the editor of the *Lancet* himself settled on the term “Down syndrome” which has continued in use more prominently to the present day.<sup>1</sup> The medically descriptive term, “trisomy 21,” which was preferred by Lejeune, is only used in the United States in reference to those cases of Down syndrome that result from full meiotic nondisjunction, and not mosaicism or translocation.

The use of language applied to those with intellectual disabilities reveals social attitudes that are often disparaging of those living with intellectual disability. The *Campaign to End the R-Word*<sup>ix</sup> (“Retard”) was launched in 2009 as a grassroots youth movement to defend those with intellectual disability from ridicule, and is evidence that discriminatory language and attitudes remain a challenge to overcome.

### **Medical Progress and Life Expectancy**

Life expectancy for those living with Down syndrome has increased dramatically from nine years of age in 1929 and 12 years in 1949 to over 60 today. A marked increase in life expectancy occurred in the 1950s with the introduction of antibiotics to fight respiratory infections which had previously been a common cause of death due to compromised immune systems exacerbated by institutionalization and the spread of infection among residents. The ability to treat cardiac defects also factors as a cause for increased longevity. Almost half of babies born with Down syndrome have congenital heart defects, the most common being atrioventricular septal defects. A recent study has shown that infants born with Down syndrome who receive surgical treatment for cardiac defects now fare better than their typically developing peers with the same condition.<sup>x</sup>

When Lejeune discovered that an extra copy of the 21st chromosome caused Down syndrome, he believed that a “cure” would one day be possible. Over 50 years later, the biological effects of trisomy 21 are much better understood, and clinical trials have even begun on potential drug therapies to improve cognition and memory, but a *cure* for Down syndrome is not considered possible.

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<sup>1</sup> For an in-depth discussion of the naming of Down syndrome, see David Wright, *Downs: The History of a Disability*.

Adding to the already acknowledged complexity of Down syndrome, an April 2014 article published in the journal *Nature* showed evidence that an extra copy of the 21<sup>st</sup> chromosome has a downstream effect on some 182 genes on chromosomes other than the 21<sup>st</sup>. The research team led by Dr. Stylianos Antonarakis at the University of Geneva Medical School had the opportunity to study gene expression of monozygotic twins, one of which had trisomy 21 and the other not. Their findings reinforced the suspected complexity of the genetic anomaly: an additional chromosome disrupts gene expression of the entire human genome, and not only the genes that reside on the 21<sup>st</sup> chromosome.

### **The Current Status of Research to Address Intellectual Disability in Those Living with Down Syndrome**

#### *Advances in Research to Improve Cognition in Down Syndrome*

The primary targets for researchers investigating treatments for Down syndrome are cognition, memory, and speech. Very recently we have seen unprecedented developments in these areas, and especially in the link between Down syndrome and Alzheimer's disease.<sup>2</sup> As of the writing of this paper, three clinical trials have been initiated by pharmaceutical companies:

- In 2011 the Roche Pharmaceutical Company began an early-stage clinical trial to investigate the safety of a drug called RG1662, and to obtain data indicating its possible effectiveness in improving cognition.<sup>xi</sup> Following a successful phase 1 trial, Roche has now initiated a phase 2 trial to evaluate the drug's efficacy in improving learning, memory, and language ability. This is an international, multi-site trial with nine clinical sites participating in the U.S.<sup>xii</sup> Interesting to note, the phase I trial involved individuals from 18 – 30 years of age. Phase 2 has lowered the recruitment age to 12 years, indicating that the initial trial showed substantial tolerability of the drug in the target population.
- In September 2013 Elan Corporation announced the first dosing of a patient in a phase 2a clinical trial with their drug, ELND005, or scyllo-inositol.<sup>xiii</sup> Scyllo-inositol is a drug thought to potentially reduce aggregation of beta-amyloid that is

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<sup>2</sup> The amyloid precursor protein (APP) gene responsible for the development of the amyloid plaques and neurofibrillary tangles that are neurological features of Alzheimer's disease is located on the 21<sup>st</sup> chromosome. All individuals with trisomy 21 have an extra copy of the APP gene and therefore will develop the neurological features of Alzheimer's disease by age 40. Almost all will experience the onset of dementia by age 60. This phenomenon has brought attention to the Down syndrome community by those working to develop treatments for Alzheimer's disease. Since it is known that all individuals with Down syndrome will develop the neurological features of Alzheimer's disease, they are the ideal control population for researchers.

- the product of the APP (amyloid precursor protein) gene located on chromosome 21. In this study, however, participants must not show evidence of dementia and the target is scyllo-inositol's effect on cognition and not memory.
- Balance Therapeutics, a new Australian pharmaceutical company, has initiated a clinical trial it is calling the “Compose Study,” for “Cognition and Memory in People with Down Syndrome”.<sup>xiv</sup> The study is investigating the safety and efficacy of a compound called BTD-001 to improve memory, language, and learning in persons with Down syndrome. BTD-001 is a form of drug that has been available since the 1920s to treat respiratory infections and dementia in the aged. The investigators have seen that the drug has potential to affect the brain signaling pathways by activating non-performing connections.

In addition, AC Immune has partnered with Genentech to identify and conduct a clinical trial on an antibody that they hope will effectively create an immune response against the development of beta amyloid.<sup>xv</sup> Other drugs are currently in the research and clinical trial pipelines, but the investment of these pharmaceutical companies is strong evidence of the current therapeutic potential for treatments to improve the lives of those living with Down syndrome.

In addition to these three industry-based clinical trials, smaller trials are also being conducted and financed privately. The Jérôme Lejeune Foundation has funded a clinical trial in Spain investigating the efficacy of a polyphenol in green tea called epigallocatechin gallate (EGCG) and its effectiveness in regulation of the DRK1A gene that is overexpressed in individuals with Down syndrome.<sup>xvi</sup> Data will be published at the end of 2014. The Jérôme Lejeune Institute in Paris is also conducting a clinical trial funded by the Jérôme Lejeune Foundation on 256 patients aged six to 18 months to investigate the efficacy of folinic acid and thyroxine hormone on improving psychomotor development of infants with Down syndrome.<sup>xvii</sup> The trial will conclude in 2017.

Researchers claim that even a decade ago young researchers were discouraged from pursuing a career investigating Down syndrome. Five years ago, clinical trials were unheard of. The accelerated pace of research in the last three to five years is a strong indication that therapeutic treatments may soon offer hope to those now living with Down syndrome and their caregivers.

### *Advances in Understanding the Neurobiology of Down Syndrome*

Researchers would be unable to develop drugs to improve the lives of those living with Down syndrome without significant developments in the understanding of the neurobiology of Down syndrome. Very recently unparalleled advances have taken place in basic science. Animal models that imitate Down syndrome, such as the



Ts65Dn mouse produced by Jackson Labs,<sup>xviii</sup> <sup>3</sup> and the ability to create induced pluripotent stem cell (iPS) lines from patients with Down syndrome have given researchers the ability to work with model systems to test their hypotheses more effectively.

Three of the more startling and recent developments in basic science will serve to illustrate the present level of achievement of researchers in advancing the understanding of Down syndrome. Two of these discoveries were hailed in the media as being a “cure” for Down syndrome, but the investigators were quick to clarify that they are not cures, but rather one more important step in understanding that may someday soon lead to therapeutic applications.

- In July 2013 Dr. Jeanne Lawrence, a researcher at the University of Massachusetts, published an article in the journal *Nature*<sup>xix</sup> showing that it is possible to silence the extra 21<sup>st</sup> chromosome completely by inserting a copy of the XIST gene (the gene which silences the X chromosome in men and the second copy of X in women) into the third copy of chromosome 21 in an iPS cell line generated from a somatic cell of a patient with Down syndrome. Hailed by the media as a “cure” for Down syndrome, Dr. Lawrence was more circumspect. She observed that “[t]he silencing of trisomy 21 by manipulation of a single gene in laboratory cells surmounts the first major obstacle to development of potential ‘chromosome therapy.’” She expressed the hope that “for individuals living with Down syndrome, this proof-of-principle (would open up) multiple exciting new avenues for studying the disorder... and bring into the realm of consideration research on the concept of ‘chromosome therapy’ in the future.”<sup>xx</sup> In the short term, what Dr. Lawrence and her lab created was a valuable tool to better understand the effect of Down syndrome on the whole genome. With the insertion of XIST, she also inserted a doxycycline “switch” that enabled her to turn on and off the extra chromosome, allowing her to test the downstream effect of an additional copy of a whole human chromosome on the entire genome.
- In September 2013 a research team led by Dr. Roger Reeves at the Johns Hopkins University School of Medicine published an article in the journal *Science*<sup>xxi</sup> showing that a single injection of a sonic hedgehog<sup>4</sup> pathway agonist at birth completely normalized cerebellar development and function of the hippocampus

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<sup>3</sup> The mouse chromosome 16 most replicates the chromosomes found on human chromosome 21. Importantly, mice are an effective, but incomplete model of trisomy 21. They do not completely reproduce the full human chromosome 21, but rather 65 genes on a critical segment of the chromosome. Other mouse strains have also been developed to investigate gene dosing in Down syndrome. Using mouse models, researchers are also able to “knock-in” or “knock-out” specific genes to study their effect in depth.

<sup>4</sup> Sonic hedgehog, first identified in fruit flies, is a protein that signals the growth of limbs, fingers, and organization of the brain in the developing fetus. It also controls the division of adult stem cells in adults and has been implicated in the development of some cancers.

in the brains of developing Ts65Dn mouse pups that lasted into adulthood, an astonishing discovery. Dr. Reeves warned that the treatment would likely have unintended effects in humans such as increasing the risk of cancer, but indicated his study highlighted the important role of the sonic hedgehog pathway in cerebellar development.<sup>xxii</sup> His results suggested a possible approach that might someday lead to therapeutic treatments to improve cognitive function in Down syndrome. Individuals with Down syndrome have a cerebellum approximately 60% the size of the typical population and reduced hippocampal function that accounts for deficits in long-term memory.

- As previously mentioned, in April 2014 a research team led by Dr. Stylianos Antonarakis at the University of Geneva Medical School in Geneva, Switzerland published their findings on a study of monozygotic twins in the journal *Nature*.<sup>xxiii</sup> In this rare set of twins, one had a full trisomy 21 and the other was typical. The most accepted theory has been that the Down syndrome phenotype is the result of the overexpression of genes on chromosome 21. These researchers proved that the perturbations exist across the entire genome and raised the question whether the Down syndrome phenotype is the result of single genes or too much DNA. If it could be discovered that it is due to single genes that regulate expression of other genes, then Down syndrome research could instantly be propelled light years ahead of where it is now. That question is next to be addressed by Dr. Antonarakis' lab.

### *Improving Birth Outcomes*

Medical research has made incredible strides toward improving the lives of those living with Down syndrome, but even more exciting are advances that have taken place in investigating prenatal therapies to improve birth outcomes.

Two research teams, one at Tufts University in Boston and the other at the University of Bologna, have published evidence that shows prenatal drug treatments improve birth outcomes in mice. Dr. Diana Bianchi's lab at Tufts has shown that fetuses with Down syndrome are subject to substantial oxidative stress in the second and third trimester of development. Her investigations into the use of antioxidant therapies delivered in utero have had astounding results in the Ts1Cje mouse.<sup>xxiv</sup> As she has stated: "It would be very exciting if prenatal screening for T21 could create an opportunity to provide fetal treatment and ultimately improve neurocognition in DS. Preliminary experiments in mouse models suggest that prenatal treatment of DS is an achievable goal."<sup>xxv</sup>

Dr. Renata Bartesaghi's lab at the University of Bologna has published on the administration of fluoxetine<sup>5</sup> prenatally from day 10 to delivery, and then giving a dose of 5-bromo-2-deoxyuridine 2 days after birth. Compared with the untreated mice, those that had received prenatal fluoxetine saw their "precursor proliferation and cellularity . . . fully restored throughout all brain regions." Additionally, "The recovery of proliferation potency and cellularity was still present in treated Ts65Dn 45-day-old mice. Moreover, embryonic treatment restored dendritic development, cortical and hippocampal synapse development and brain volume. Importantly, these effects were accompanied by recovery of behavioral performance."<sup>xxvi</sup> Of substantial concern, however, is the safety of this particular drug, especially when administered to human fetuses in utero.

The particular significance of these studies is that they show the very real potential that exists for reversing the neurodevelopmental deficits associated with Down syndrome that begin during fetal development. Success in this particular area of research could have a transformative effect on the decision parents face following a prenatal diagnosis of Down syndrome, a decision that now strongly favors abortion.

### *Scant Funding*

A significant impediment to more rapid progress in identifying treatments for Down syndrome has been funding. The National Institutes of Health (NIH) has received harsh criticism from the Down syndrome community for their meager spending on Down syndrome in their extramural research portfolio. A mere \$22 million was appropriated in 2010, but in the 2014 and 2015 (est.) budgets, \$19 million will be spent on Down syndrome research.<sup>xxvii</sup> If that allocation is compared to other disabilities on a per capita basis, Down syndrome receives a fraction of support. The comparison commonly cited is with research for cystic fibrosis. \$80 million has been allocated for 2014/2015 for a population of approximately 30,000 people. That is approximately \$2,666 per person as compared to \$76 per person for Down syndrome using a population of 250,000 for the calculation. The most common genetic cause of intellectual disability is one of the worst funded in the NIH research portfolio. The burden of funding, then, is with private foundations that rely on the generosity of their donors to support research that will improve the lives of a significant number of the population.

In spite of low funding levels, in recent years the National Institute of Child Health and Human Development (NICHD) at the NIH under the leadership of Dr. Yvonne Maddox has shown tremendous support for Down syndrome research and care. Dr. Maddox Initiated the NIH Down Syndrome Consortium to gather together

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<sup>5</sup> Fluoxetine is commonly known by the trade name Prozac.

representatives from the Down syndrome research and advocacy communities, and with Consortium assistance she has launched DS-Connect, an online contact registry to which individuals living with Down syndrome can subscribe to be notified of upcoming clinical trials.

## Prenatal Diagnosis and Abortion

### *Historical Context*

Prenatal diagnosis and selective abortion have emerged since the 1970s as two sides of the same coin. The technique of creating a karyotype from human cells to identify Down syndrome was first developed in France and published by Lejeune, Gautier and Turpin in 1959. In 1968 it was discovered that fetal cells in amniotic fluid could be used to generate a karyotype that would identify Down syndrome as early as the 16<sup>th</sup> week of pregnancy. With that discovery, social attitudes toward Down syndrome rapidly shifted toward seeing it as something that could, and often should, be avoided. Following the U.S. Supreme Court 1973 decision in *Roe v. Wade*, which created a constitutional right to abortion, some began to question whether society had an obligation to use abortion to purge the human race of disease and disability.

From the beginning of the debate over prenatal diagnosis and abortion, Down syndrome has been a focal point of competing arguments over how we view and respond to disability. Harry Harris, in his 1974 book, *Prenatal Diagnosis and Selective Abortion*,<sup>xxviii</sup> said that prenatal diagnosis had provided a new objective in medicine, and that the new objective was not the nature of the technique, but rather the objective at which the diagnosis is aimed. He went on that the object of prenatal diagnosis is “to find out whether the foetus has some defined abnormality which will inevitably lead to the birth of a defective infant and, if so, to abort the foetus.” Of course, the example of an “abnormality” that he used in his argument was “mongolism.” He mentioned Jérôme Lejeune’s opposition to abortion on the grounds that it was unethical and immoral, but continued to say that most human geneticists disagreed with Lejeune’s position because it “dismisses too easily the welfare of afflicted families and the general social good.”

Far from an isolated perspective, Harris’s view was promoted by Dr. Joycelyn Elders, who became the U.S. Surgeon General during the Clinton administration. Dr. Elders was the first public figure to advocate for the abortion of Down syndrome babies. She was quoted in 1990, when she was Arkansas State Health Director, that “[a]bortion has had an important, and positive, public-health effect” because it has reduced “the number of children afflicted with severe defects.” To provide an example to support her claim, she stated, “The number of Down’s syndrome infants

in Washington State in 1976 was 64 percent lower than it would have been without legal abortion."<sup>xxix</sup>

Elders' position, it could be argued, has become enshrined as a best practice in medical genetics and obstetric care. A poll of the members of the American College of Obstetricians and Gynecologists (ACOG) conducted in 1995 revealed that 63% of the members who responded believed abortion was a justifiable *treatment option* for fetal anomalies compatible with life. Ninety percent believed abortion was justifiable for uniformly fatal fetal anomalies.<sup>xxx</sup> Even more recently, a 2013 poll, the results of which were published in the *Journal of Intellectual and Developmental Disabilities*, showed that almost one in four patients who had received a positive prenatal diagnosis for Down syndrome said their medical professional was *insistent* that they terminate their pregnancy.<sup>xxxi</sup> ACOG now recommends that all women, regardless of maternal age, be offered prenatal screening for aneuploidy (trisomies), either by screenings or invasive testing such as amniocentesis. This recommendation, combined with the prevailing attitude toward birth defects in the medical community, would seem to encourage the option for abortion following a positive result. As the now-deceased disabilities rights activist, Adrienne Asch, has stated, prenatal diagnosis "is not a medical procedure to promote the health of the fetus. It is a procedure to give prospective parents information to decide whether or not to eliminate a possible future life."<sup>xxxii</sup>

### *Defining Abortion Statistics*

Determining what this information portends in terms of the actual number of abortions of Down syndrome pregnancies in the U.S. is difficult. The most commonly recited abortion statistic in the U.S. is 92% termination following a positive prenatal diagnosis for Down syndrome; however, it is not likely an accurate statistic. This number is derived from a study published in 1999 that reviewed 10 studies on Down syndrome published between 1980 and 1998.<sup>xxxiii</sup> Only three of those studies were from the United States; the most recent was from 1988 and comprised only 77 of the 5,035 patient cases reviewed.

In 2012 a new review of the literature on abortion following prenatal diagnosis was published.<sup>xxxiv</sup> After applying a rigid exclusion criterion to 308 potential publications, 24 articles were accepted by the authors to include in their review, including population-based, hospital-based, and anomaly-based studies. Their analysis of the literature showed that termination rates following prenatal diagnosis vary according to a number of factors such as maternal age, race and ethnicity, and gestational age, but the range averages from 67% - 85%.<sup>6</sup> Of particular interest,

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<sup>6</sup> See the publication for a breakdown of statistics by each study category.

however, is the finer analysis of the study. In hospital-based studies, they noted that one publication showed higher termination rates associated with earlier gestational age, i.e., 93% at 16 weeks gestation or less, as compared to 85% at 17 weeks or greater. The implication of this finding is of particular concern given that the newest methods of prenatal screening are able to return a result as early as 10 weeks of gestation,<sup>7</sup> well within the first trimester, and at a point in pregnancy when there may be less maternal attachment and inhibition regarding termination.

As evidence of the ambiguity of abortion statistics following prenatal diagnosis, the Natoli study revealed some positive trends. Compared to the earlier Mansfield study, Natoli, et al. suggested that selective abortion for Down syndrome in the U.S. declined through the 1990s and early 2000s. The authors of another study, however, offer a differently nuanced view. They claim that the Down syndrome birthrate declined sharply following passage of the *Americans with Disabilities Act* in 1990, and only began to rebound in 1995 and almost reach its previous level by 2002.<sup>xxxv</sup> Possible reasons for this decline and reemergence of Down syndrome births will be discussed in a later section.

Of note, the authors of the Natoli study present analyses of data from 20 registries in the International Clearinghouse for Birth Defects Surveillance and Research that indicate abortion rates after Down syndrome diagnosis in North America are lower than in Europe and Australia.<sup>xxxvi</sup> Perhaps U.S. advocacy efforts to improve the quality of life and acceptance of those living with Down syndrome have been successful. One can only hope this trend will continue and not be offset by the availability of early prenatal diagnosis using new and sophisticated noninvasive prenatal screening tests.

Dr. Brian Skotko has given an overall impression of what these statistics mean with regard to the Down syndrome population in the United States and abroad. His research shows that due to prenatal testing, “[T]he worldwide birth incidence of DS has actually decreased from what it could have been by 2–18% per year,” and that “in the USA, there would have been a 34% increase in the number of babies born with DS between 1989 and 2005, in the absence of prenatal testing. Instead, there were 15% fewer babies born, representing a 49% decrease between the expected and observed rates.”<sup>xxxvii</sup> Indeed, in 2008 the population statistics of those living with Down syndrome in the United States were revised downward from 400,000 to 250,000.<sup>xxxviii</sup>

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<sup>7</sup> These include so-called “noninvasive prenatal screening tests” (NIPS) that identify cell-free DNA from the fetus in the mother’s blood and then subject it to microarray analysis for trisomies 21, 18, 13, and some X-linked chromosomal disorders. These technologies are heavily promoted by their manufacturers - Sequenom (MaterniT21), Ariosa (Harmony), and Illumina/Verinata (Panorama) - and they claim a sensitivity range between 99.1% and 99.9% with very low false positive rates.

Whatever statistics one might choose to cite, it is clear that the majority of women who receive a prenatal diagnosis of Down syndrome choose to terminate their pregnancy. Whether the number is 50%, or 93%, it is a staggering social problem reflective of society's attitudes toward and perception of disability in general, specifically intellectual disability, and especially in a category of disability so easily targeted. Those living with Down syndrome cannot hide their disability; they wear it on their faces and are easy targets for fear and discrimination.

*“Wrongful Birth”*

Fueling concerns about the future of expanded prenatal testing, a number of high-profile wrongful birth lawsuits have succeeded in U.S. courts and abroad.

In 2012 a Portland, Oregon couple was awarded \$2.9 million in a court case in which they claimed that they would not have allowed their child to be born had they known she would have Down syndrome.<sup>xxxix</sup> They claimed that the doctors were “negligent” in their prenatal care. The family had initially sued for \$7 million to cover the cost of providing for their child over the course of her lifetime.

In December 2013 a jury awarded another couple \$50 million when they claimed that had they known their child would be born with a severe genetic defect, an unbalanced chromosomal translocation, they would have chosen to abort.<sup>xl</sup> They have sued both the medical practice and Laboratory Corporation of America, the company they claim missed the translocation in reading the test.

The success of these wrongful birth lawsuits relates back to the legal precedent established in the New York courts in 1978. In the case, *Becker v. Schwartz*, the couple claimed to have not been adequately informed of the risk for Down syndrome in older mothers nor offered an amniocentesis.<sup>xli</sup> For the first time, a court acknowledged hardship imposed by medical negligence in failing to provide an opportunity to abort a child, and awarded the cost of raising their child through its lifetime.

The potential impact of these wrongful birth lawsuits on influencing prenatal testing guidelines cannot be overlooked. If doctors can be held liable for not offering prenatal diagnosis, or for “missing” a genetic anomaly, then risk exposure for insurance companies is increased and prenatal testing becomes free and encouraged. Important questions are then raised regarding a physician's freedom to practice medicine according to conscience, prenatal diagnosis for the purpose of abortion becomes a right, and contemporary expectations of childbirth lead to parents' increasingly presumed right to choose which babies are allowed to come

into the world. The way that these questions are resolved around Down syndrome now will prepare the ground for management of future cases as prenatal diagnosis becomes increasingly sophisticated, less expensive, and more broadly and simply available.

In the short time since noninvasive prenatal screening tests have been commercially available (October 2011), the tests have been expanded to the point where manufacturers now claim the ability to identify not just aneuploidy, but also DiGeorge, cri-du-chat, Prader-Willi/Angelman, and 1p36 deletion syndromes.<sup>xlii</sup> Advanced microarray testing that will be able to do whole genome genetic analysis of fetal cells in maternal blood is in development. These tests could potentially replace amniocentesis and chorionic villus sampling, both of which carry a risk of miscarriage, and safely identify at nine weeks into a pregnancy most known genetic abnormalities.<sup>8</sup>

Given the success of litigation for wrongful birth recommendation of prenatal testing for all women by ACOG, provision for prenatal testing as part of the “Women’s Preventive Services Guidelines” contained under the “Affordable Care Act,”<sup>9</sup> and the evident self-interest insurance companies have in reducing exposure to the financial liability of providing coverage for the birth of a medically challenged child, it is critical that protections be put into place for children who are emotionally victimized by their parents’ legal complaints that they did not have the option to abort them. Tort reform efforts should be initiated to limit incentives for these persons to sue their health care providers.

Rep. Stephen Palazzo (R) from Mississippi introduced such legislation, H.R. 4698, on May 21, 2014 with the intent to prohibit certain wrongful birth and wrongful life civil actions.<sup>xliii</sup>

### ***Attempted Legal Protections of the Unborn with Disabilities***

The landmark disabilities legislation in the United States is called the *Americans with Disabilities Act* (ADA). The act was signed into law by President George H. W. Bush in 1990 and then broadened by President George W. Bush when he signed into law the ADA Amendments Act of 2008. The ADA was intended to have the same effect of ending discrimination based upon disability as the Civil Rights Act of 1964

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<sup>8</sup> Cell Scape Corporation is one company developing a noninvasive test they call “Clarity.” See detailed information at their website, <http://www.cellscapecorp.com/>.

<sup>9</sup> Under the Final Rules of the “Women’s Preventative Services Guidelines,” recommendations supported by the Health Resources and Services Administration that are not specifically mentioned in the Guidelines must be covered without any cost-sharing requirement.



had for racial minorities.<sup>xliv</sup><sup>10</sup> The ADA reflects important truths about the dignity of disabled persons that can also be invoked in the context of opposing the practice of disability-based abortion.

Lawmakers are beginning to address the issue of disability-selective abortion. A bill banning disability-selective abortions was signed into law in North Dakota on March 26, 2013. After challenging the law in the courts, the Fargo-based Red River Women's Clinic, with assistance from the Center for Reproductive Rights, dropped their opposition without prejudice to the provision to ban abortions because of sex or disability citing that they don't perform abortions for those purposes anyway. The bill had also attempted to ban all abortions after six weeks. That six-week ban was blocked by a federal district court judge who called it "clearly invalid and unconstitutional."<sup>xlv</sup>

Similar legislation failed earlier this year in South Dakota where a ban on both sex-selective and disability-selective abortions was attempted in 2014. HB 1240 failed which would have prohibited disability-selective abortions, but the ban on sex-selective abortions was sustained.

Legislation was also introduced in Indiana in the 2013 legislative session that would have prohibited sex-selective and disability-selective abortions. The legislation would have made the knowing provision of an abortion for these reasons a Class-C felony in the state but the bill died in committee.

In a brief filed on behalf of the Jérôme Lejeune Foundation, Saving Downs, and the International Down Syndrome Coalition, the Bioethics Defense Fund has argued that the U.S. Supreme Court "has never endorsed a right to abort children only because they have been detected to have a disability." In the 1992 *Planned Parenthood v. Casey* decision, the Bioethics Defense Fund brief argues, the Supreme Court "repeatedly premised its reaffirmation of abortion rights in terms of the right to terminate an unintended pregnancy . . . . [The Supreme] Court has never framed the protected abortion decision as whether to bear or abort a *particular* child based on

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<sup>10</sup> It has been suggested by some authors that the immediate effect of the ADA was, at least with consideration of abortion, an increase in termination rates following prenatal diagnosis. Fox and Griffin suggest that there were two consequences of the ADA. Immediately after passage, they claim it may have "promoted disability-selective abortion if social interactions reinforced negative attitudes toward people with disabilities, or if the media portrayed people with disabilities as incurring undesirable costs for society. Their data suggests that with no increase in prenatal diagnosis, birth rates fell from 1989 to 1995 by between 13 and 18 per 100,000 births, adding controls for other demographic and medical variables. This information is provided only to suggest another parallel between civil rights legislation and the ADA, and that is that public attitudes are not necessarily changed with protective legislation. In the years between 1998 and 2002 birth rates of children with disabilities increased, but did not reach their pre-ADA levels.

identified traits of genetic variation, disability, or other health condition.” According to the Bioethics Defense Fund brief, the Court’s decision in *Casey* “formulated the abortion decision as one confronting a woman ‘when the woman confronts the reality that, despite her attempts to avoid it, she has become pregnant’ – not when she accepts a pregnancy at first, but then comes to perceive the child she is carrying as defective”<sup>xlvi</sup>

### *Down Syndrome Prenatal Education – Legislation*

The link between prenatal testing and abortion raises significant questions around informed consent that have not yet been adequately addressed, but that are important to ensuring that families are aware of the liabilities and benefits of testing, and certainly before they act upon it to end the life of their child. Adrienne Asch and David Wasserman, writing in the “Virtual Mentor,” a publication of the American Medical Association, outlined those issues as follows:

1. When is the best time to introduce the subject of testing;
2. What type of information about the tests do prospective parents want or need;
3. What is the proper balance between medical information and information on nonmedical aspects of life with a particular disease or disability;
4. How can the perspectives of people living with the conditions and their families best be included; and
5. How can uncertainty about the applicability of general information to a specific child and family situation be conveyed?<sup>xlvi</sup>

This article by Asch and Wasserman makes several excellent observations that can provide talking points and inspiration for those working to pass legislation that protects families and secures the safety of children prenatally diagnosed with a disability. Research shows information now supplied following prenatal diagnosis is biased, outdated, narrow, inaccurate and clinical<sup>xlvi</sup>; and, as has been previously stated, this information comes from a profession which prefers termination in the case of disability. Informed consent give parents an opportunity to look on their child as an individual person living with a disability, and not as a person who will be consumed with a disability. Studies show that helping parents see disability in the context of a full human life will influence their response to a prenatal diagnosis.<sup>xlvi</sup>

Acknowledging that the way prenatal testing is offered and the results explained do not adequately ensure informed consent, Senator Sam Brownback and the late Senator Ted Kennedy introduced into the U.S. Senate the *Prenatally and Postnatally Diagnosed Conditions Awareness Act* in 2007. The act was intended to “increase the provision of scientifically sound information and support services to patients receiving a positive diagnostic test for Down syndrome, or other prenatally or

postnatally diagnosed conditions.”<sup>1</sup> The act was passed by the Congress and signed into law by President George W. Bush on October 8, 2008. Unfortunately, the bill has not been funded due to disagreement between prolife and prochoice legislators over how the topic of abortion would be handled in the materials accepted for distribution.

Advocates view the Kennedy-Brownback bill as a good first step, but one that falls short of addressing a key moment, and that is before the test is offered to the patient, providing information on potential outcomes in advance of a result. Prenatal screening tests are most often given as part of routine prenatal care, as recommended by ACOG, with no understanding by women of potential outcomes, or even the purpose for which the tests are being offered. An unanticipated result can place women in a position of vulnerability to the influence of their health care provider. Asch and Wasserman provide evidence that many women would decline prenatal testing altogether if they were adequately informed before the test is given of the rewards and challenges of raising a child with a disability.

In the absence of funding for the Kennedy-Brownback bill, advocates in various states have pursued legislation with the same intent. To date, six states have passed their own versions of prenatal education legislation intended specifically to require information regarding the positive outcomes of giving birth to a child with Down syndrome. Those states are: Virginia (Virginia Code 54.1-2403.01.B), Missouri (Revised Statutes, 191.923), Massachusetts (H 3815, 2012), Kentucky (SB 34, 2013), Delaware (HB 214, 2014), and Maryland (SB 654, 2014). Legislation is currently pending in the following states:

- Louisiana (H.B. 1058). Passed both houses of legislature on May 20, 2014 and awaiting governor’s signature
- New Jersey (A 3233). Introduced May 22, 2014
- Ohio (HB 552). Introduced May 19, 2014
- Oklahoma (SB 586, 2013). Introduced Feb. 5, 2013, referred to committee
- Pennsylvania (HB 2111/SB 1339). Passed in the House of Representatives on May 6, 2014, and awaiting vote in Senate

The Louisiana legislation (H.B. 1058) introduced in 2014 includes a unique and controversial element. H.B. 1058 requires that all resource materials provided to women not mention abortion as an option following a positive prenatal diagnosis for Down syndrome. Using a prenatal discrimination argument, the language of the bill states that information must be provided that “[d]oes 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.” Opponents argue that states cannot limit information on legally permissible procedures. This provision was signed into law by Governor Bobby Jindal on May 30, 2014 and is nearly certain to be challenged in court.<sup>li</sup>

The National Down Syndrome Society (NDSS) provides a “Prenatal Information State Law Toolkit” for those wishing to propose legislation in their states. The toolkit includes model legislation, sample press releases, and samples of testimonies.<sup>lii</sup>

### *Down Syndrome Prenatal Education – Resources*

States continue to pass laws nationwide which require the provision of a list of peer supports and up-to-date, evidence-based, written information to those who receive a prenatal diagnosis of Down syndrome. However, the currently available resources are few.

The most widely available and well-presented resource is *Understanding a Down Syndrome Diagnosis*, available at [www.lettercase.org](http://www.lettercase.org) and from the National Center for Prenatal and Postnatal Down Syndrome Resources of the Human Development Institute at the University of Kentucky. The preparation of this small booklet was assisted by representatives from the National Society of Genetic Counselors, the American College of Medical Genetics, ACOG, the National Down Syndrome Society, and the National Down Syndrome Congress. With those endorsements, it qualifies under the emerging legislation as a peer-reviewed resource. Critics of the *Lettercase Booklet*, as it is commonly called, question its acknowledgement of abortion as a choice many families make. The primary authors defend that decision, stating that it is the only way they could have obtained the endorsement of the medical community that is essential in promoting distribution to doctors and genetic counselors who will then place it in the hands of those receiving a prenatal diagnosis of Down syndrome. The small section on “Pregnancy Termination” is intentionally placed in counterpoint to the much larger section on “Adoption” which appears on the opposing page.

The National Center also publishes *Diagnosis to Delivery: A Pregnant Mother’s Guide to Down Syndrome*, and will soon have available *Coping with Loss* for parents who have lost their pregnancy or newborn child. The Center also maintains two web sites in addition to their portal at [www.DownSyndromeDiagnosis.org](http://www.DownSyndromeDiagnosis.org): one site is [www.BrighterTomorrows.org](http://www.BrighterTomorrows.org), which provides comprehensive resources for expectant parents and training modules for physicians, and the other is [www.DownSyndromePregnancy.org](http://www.DownSyndromePregnancy.org), a website with a blog for women who are pregnant and expecting a baby with Down syndrome. All resources from the

National Center for Prenatal and Postnatal Down Syndrome Resources are available in both English and Spanish.

In 2012 the Global Down Syndrome Foundation and the National Down Syndrome Congress together published a small pamphlet, available in print and online at [www.downsyndrometest.org](http://www.downsyndrometest.org), that they hoped would “eliminate confusion at a time when advice and guidance can vary dramatically from doctor to doctor and counselor to counselor, and where there was previously no accessible, consistent resource for women and families.”<sup>11</sup> The authors have stated that self-advocates<sup>11</sup> resent abortion being mentioned in resources provided to women who have received a prenatal diagnosis for Down syndrome; consequently, this resource makes no mention of abortion as an option following prenatal diagnosis. The sponsoring organizations initially arranged for Sequenom Center for Molecular Medicine, the first company to make commercially available a prenatal screening test to identify aneuploidy in cell-free DNA in maternal blood in October 2011, to share printing costs and distribute the pamphlet along with orders for the MaterniT21 test. The current status of that arrangement is not known.

In addition to these two print and online resources, various organizations provide informational support for women who have received a prenatal diagnosis of Down syndrome. Be Not Afraid ([www.benotafraid.net](http://www.benotafraid.net)) is an organization whose ethical principles are informed by the teachings of the Catholic Church. They provide a referral service that connects parents with a peer minister for those who have received an unexpected prenatal diagnosis. Other organizations provide online information and support such as the Down Syndrome Diagnostic Network (<http://www.dsdiagnosisnetwork.org>), the International Down Syndrome Coalition ([www.theidsc.org](http://www.theidsc.org)), and others available online. Many individuals who staff these organizations consider themselves prolife, but prefer to be seen as “pro-information” and use “non-directive” language in counseling those who contact them to engage those who may be considering termination. Individuals who believe that prenatal counseling should discourage abortion have criticized this “non-directive” approach. Counselors, however, defend their approach by saying that women considering abortion will avoid any resource they believe is attempting to deny them the option to abort.

### *The Education and Training of Health Care Professionals*

In the aforementioned study on parental experiences in receiving a Down syndrome diagnosis (Goff), negative experiences outnumbered positive experiences 2.5 to 1.

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<sup>11</sup> Self-advocates are individuals living with Down syndrome who speak publicly to raise awareness of their disability and to show the capabilities of those living with Down syndrome.

The reasons listed for these negative experiences included the health care provider's insistence on termination, the communication of negative stereotypes of individuals with Down syndrome, a lack of available information, and a perceived lack of compassion shown by the physician and staff.<sup>liv</sup> These findings were not surprising. In a survey conducted by the Special Olympics, 81% of medical school students said they do not get any clinical training regarding individuals with intellectual disabilities.<sup>lv</sup> Forty-five percent of ACOG fellows and juniors reported their residency training as "barely adequate or nonexistent," and only 28% of ACOG fellows felt "well qualified" to provide genetic counseling.<sup>lvi</sup>

The response of medical professionals to those who receive an unanticipated prenatal diagnosis of Down syndrome is key to the decision women make regarding birth or abortion. In a 2007 study done in the Netherlands, 97% of women who chose to terminate their pregnancy following a prenatal diagnosis of Down syndrome claimed they had chosen to abort a baby they had wanted until receiving the diagnosis. Their motivations for termination were based on misunderstandings of the disability and included as reasons: a belief the child would never be able to function independently (92%), a consideration that the abnormality was too severe (90%), the burden for the child himself of having the disability would be too heavy (83%), uncertainty about the consequences of the disability were too great (78%), and the burden for other children in the family would be too heavy (73%).<sup>lvii</sup>

These reasons given for termination lie in stark contrast to the real, lived experiences of families who have accepted a child with Down syndrome into their homes. A summary of those statistics was provided at the beginning of this paper. These reasons reveal a perception of Down syndrome that is uninformed by the reality of the lived experiences of thousands of families – a reality those professionally bound to the principle of informed consent should be obligated to communicate.

The incidence of abortion of children with Down syndrome is inseparably tied to the promotion of prenatal screening. If the statistics on the number of abortions following a prenatal diagnosis of Down syndrome is to change, five key elements must be addressed with regard to the delivery of the diagnosis and care of patients:

1. Standardized practice guidelines should be promulgated among medical professionals regarding how best to deliver a prenatal and postnatal diagnosis of Down syndrome and other genetic intellectual disabilities.<sup>12</sup>

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<sup>12</sup> Practice guidelines have been developed by The National Society of Genetic Counselors and are available at their website at [www.nsgc.org](http://www.nsgc.org). The guidelines reinforce the principles expressed in the 2008 Kennedy–Brownback bill with regard to balancing positive and negative outcomes, but are insistent that "feelings about having a child with Down syndrome dictate the conversation that

2. Accrediting agencies must require that health care professionals, as part of their medical education, be trained in how to deliver complete and consistent information free of personal bias against disabilities and to care for individuals with intellectual and developmental disabilities.
3. Prenatal information resources, similar to those listed above, must continue to be developed that are linguistically and culturally appropriate.
4. Legislation should be enacted and enforced nationwide requiring that when women are given a prenatal diagnosis of Down syndrome, at the same time they are provided a list of peer-support organizations in their area, and accurate, up-to-date, evidence-based information on the outcomes of delivering a child with Down syndrome.
5. Public education efforts must continue regarding the value of individuals living with Down syndrome.

Research has confirmed that the “overall impression of the families and children with Down syndrome is one of normality.”<sup>lviii</sup> However, historically research has focused on the negative aspects, or a “deficit framework” of disability as a “life stressor.”<sup>lix</sup> The challenge advocates face is restructuring the framework of disability to one of acceptance and love through the provision of accurate information, and the experience of families who find that having a child with a disability increases their mutual love and sense of resilience.

## **Conclusion**

This paper began and will conclude by pointing out the discrepancy between experiences of those living with Down syndrome and their families, and the negative stereotype of Down syndrome that is still deeply embedded in culture. That stereotype leads to the harshest form of discrimination against individuals with Down syndrome: selective abortion following prenatal diagnosis in a majority of cases.

The Down syndrome community is an incredibly diverse one with a wide range of abilities and disabilities. Families who live with Down syndrome and those who have no experience with Down syndrome are more similar than different. Public and many private schools welcome children with Down syndrome; they are able to learn to their capacity; they are often employed; and some now attend special college programs, live independently or in group homes, and even marry.

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follows regarding options,” which include termination, and that nondirective, non-judgmental language be used in counseling. The stated purpose of the guidelines is to “uphold patient autonomy regarding reproductive choices.” Understanding the radical and destructive choice that is abortion, the guidelines could have benefited by less neutrality and more opportunity to introduce couples to families who have children with Down syndrome.

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So what can be done in the face of increasingly sophisticated means of prenatal diagnosis, and as parents are offered the opportunity to become increasingly selective of the lives they accept into their families? This paper has attempted to show that getting accurate information to women and their partners is key in the near term if we are to rescue from abortion those prenatally diagnosed with a disability.

Jérôme Lejeune, however, would have said that “the only way to save them is to cure them.” He believed that research would one day resolve the metabolic disorder caused by an extra 21<sup>st</sup> chromosome and remove the fear and stigma of Down syndrome in the minds of parents and the community. It has been shown in this paper that research has made incredible progress toward that goal in a short period of time. Those living with Down syndrome have mild to moderate intellectual disability. It may soon be possible to improve cognition in those living with Down syndrome enough to ensure employment and independence for many. It may also soon be possible to restore neurological development before birth, radically changing even the best story that can now be given to women who receive an unexpected prenatal diagnosis of Down syndrome. It is critical that NIH funding levels be increased to support science that is just now translating into therapeutic trials to improve the lives of those living with Down syndrome.

Abortion is not the preference of families that receive a prenatal diagnosis of Down syndrome, but is chosen because of fear of an uncertain future, grief over the loss of an image parents had in mind for their child and their family, concern that their child will suffer, concern over a lifetime of managing health issues, and other similar concerns, most of which can be dispelled by the experiences of families living with Down syndrome. The value of peer support following prenatal diagnosis cannot be overestimated. As one parent commented in the Goff study, “Talking to parents of kids with DS and meeting beautiful children with DS helped us to be comforted that our son would be just like any other child.”<sup>ix</sup>

The lives of those living with Down syndrome have been improved radically since the genetic cause was discovered in 1958. The challenge remains, however, to use legislative means to break through barriers to communication, often created by the medical community, so that quality of life, both for the individual and the family, is communicated in an accurate and effective way. By this means this final and most deadly bastion of discrimination can fade into the past along with the institutions which once housed these children to keep them far from the general population.

Summary of Recommendations



1. Funding and enforcement of the Kennedy-Brownback *Prenatally and Postnatally Diagnosed Conditions Awareness Act of 2008* should be a top priority. Concern over the possibility of information being provided that mentions abortion should not impede passage of legislation that will place in women's hands information on positive outcomes. As this paper has shown, research supports the claim that when positive information is provided with appropriate peer supports, the incidence of abortion is reduced.
2. In the absence of funding for Kennedy-Brownback, advocates should work at the state level to pass legislation similar to that which has been passed in other states mentioned in this paper. Each state's sensitivity regarding abortion should be considered in proposing legislation. The desire to include language or provisions that discourage abortion should not place at risk passage of legislation that would be a positive force toward reducing the incidence of abortion following prenatal diagnosis.
3. NIH funding levels for Down syndrome research should at least equal funding for similar intellectual disabilities. Fragile X syndrome is the most common form of inherited intellectual disability and could serve as a first benchmark to increase funding for Down syndrome. The number of people living with fragile X is unknown, so a *per capita* suggested level cannot be given. Funding for fragile X syndrome, however, has increased since 2010 from \$25 million to \$30 million while funding for Down syndrome has decreased from \$22 million to \$19 million.<sup>lxi</sup>
4. Advocates should introduce and advocate for prenatal nondiscrimination legislation in states that would protect children prenatally diagnosed with Down syndrome from abortion. The argumentation in the amicus brief presented to the Supreme Court by the Bioethics Defense Fund (cited above) on behalf of the Jérôme Lejeune Foundation and two other advocacy organizations can serve as a model for arguing the legitimacy of such legislation. Americans United for Life also provides model legislation that can be used by those wishing to introduce legislation banning disability-selective abortion.<sup>lxii</sup>
5. Support should be given for tort reform legislation proposed by Rep. Stephen Palazzo, or other forthcoming legislation, that limits liabilities of physicians in wrongful birth lawsuits.

*\*Mark Bradford is President of the Lejeune Foundation USA.*

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<sup>i</sup> U.S. Centers for Disease Control, "Facts about Down Syndrome," <http://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>

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- <sup>xiii</sup> See, <http://www.clinicaltrials.gov/ct2/show/NCT01791725?term=%22down+syndrome%22+elan&rank=1>
- <sup>xiv</sup> See <http://compose21.com/study.htm>
- <sup>xv</sup> Scrip Intelligence, “AC Immune and Genentech partner on first-in-class Alzheimer’s antibody.” Accessed online, June 4, 2014 at [http://acimmune.com/content/img/pages/20120619\\_ScripReprint.pdf](http://acimmune.com/content/img/pages/20120619_ScripReprint.pdf)
- <sup>xvi</sup> See: <http://clinicaltrials.gov/ct2/show/NCT01699711?term=EGCG&cond=%22Down+Syndrome%22&rank=2> and <http://lejeuneusa.org/node/563#.U6sSKY1dXRI>
- <sup>xvii</sup> See <http://clinicaltrials.gov/ct2/show/NCT01576705?term=jerome+lejeune&rank=4> and <http://lejeuneusa.org/node/562#.U6sSgo1dXRI>
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