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To the Distinguished Chair and Honored Members of the Committee.

Thank you for the opportunity to testify on SB 1457, the bill to prohibit prenatal discrimination, by prohibiting abortion based on diagnosed or suspected genetic abnormality.

I am a cell and developmental biologist, and have over 40 years experience as a scientist, researcher and professor. My scientific experience includes federally-funded laboratory research, including post-doctoral work at Los Alamos National Laboratory, and past academic appointments at the University of Texas Medical School-Houston, Indiana State University and Indiana University School of Medicine. My professional experience includes scientific research, teaching and mentoring graduate and undergraduate students, medical and nursing students, in the areas of cell biology, embryology and developmental biology, cell and tissue culture, molecular biology and biochemistry. I am testifying in my capacity as a scientist, at the request of Sen. Barto.

This bill deals with preventing discrimination based on genetic differences, in pre-born human beings. While it might seem to some people that this is a straightforward and logical protection that is unnecessary, there is ample evidence for the need of such protection.

The human genetic composition is determined at the moment of conception, including genetically-determined traits such as gender (determined by the sex chromosome composition), hair color, eye color, and other human traits. Likewise many genetic abnormalities, such as Down syndrome in which an individual has an additional chromosome 21, Edwards syndrome which is trisomy 18, Patau syndrome which is trisomy 13, DiGeorge syndrome (22q11.2 deletion) and numerous other chromosomal, single-gene and multi-gene anomalies are determined at conception when the sperm and egg fuse to form the zygote, the single-celled human organism.

For the purposes of this legislation: “genetic abnormality” means the presence or presumed presence of an abnormal feature in an unborn child’s genome, including a chromosomal disorder or morphological malformation often caused by aberrant gene expression.

Eugenics is the term given to attempts to control human heredity. In the past, such attempts have included efforts at selective breeding of “high-quality” individuals, selective sterilization of others to prevent offspring, and even infanticide. Today we see eugenic selection attempts through abortion, what has sometimes been termed “selecting against” certain traits or individuals deemed undesirable.

How are genetic abnormalities detected in the developing fetus? Ultrasounds are often one of the first indications, as well as the Quad Screen Test, a maternal blood test that looks at circulating

levels of a group of four hormones and proteins. The actual diagnostic test, especially in the case of trisomies such as Down syndrome, is to directly visualize the chromosomes of the developing baby, done by obtaining cells from the baby or amniotic fluid through the techniques of amniocentesis or chorionic villus sampling. However, the use of Non-Invasive Prenatal Tests (NIPT), in which a sample of maternal blood is taken and then tested for genetics of the unborn child based on fetal DNA present in the blood, are beginning to supplant chromosomal tests via amniocentesis or chorionic villus sampling.[1] These tests have been refined such that they can now detect, even earlier than the invasive techniques, not only chromosomal trisomies but also other chromosomal and genetic changes, with an increasing list of genetic disorders and traits that can be identified.[2,3] The technique has the ability to detect abnormalities on a genome-wide basis,[4] and even to sequence the entire fetal DNA genome.[5,6]

Genetic discrimination abortions, destroying individuals with genetic abnormalities or suspected genetic abnormalities, show well-documented evidence involving discrimination against babies diagnosed *in utero* with Down syndrome as well as other genetic differences. Studies show that children diagnosed in the womb with the genetic characteristics of Down syndrome –trisomy 21, i.e., having 3 copies of chromosome 21 – are aborted at an extremely high rate. Documentation from other countries, which keep better records than the United States, tells a chilling tale.

In the U.K., an earlier study found a 92% abortion rate for children diagnosed in the womb with Down syndrome,[7] while a study first published in 2015 found that 74.4% of babies in the womb detected with Down syndrome using NIPT, and 82.1% of babies detected with Trisomy 13 or 18, were aborted.[8] Maxwell and co-workers reported a 93% abortion rate in Western Australia for babies diagnosed in the womb with Down syndrome.[9] De Graaf and colleagues looked at the Down syndrome population throughout Europe and found that there were 50% fewer babies born with

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Down syndrome looking back 40 years up to 2015, and that just over the period of 2011-2015, abortions decreased the Down syndrome population in Europe by a rate of 27%.[10,11]

In 2009, Skotko posed the question of whether the new non-invasive prenatal testing would mean babies with Down syndrome would slowly disappear.[12] In less than a decade, his question was answered. In 2017, Iceland reported that it was on pace to virtually eliminate Down syndrome through abortion.[13] Denmark was the first country to institute a national screening program, and has seen Down syndrome births drop dramatically.[14] Denmark is moving closely on the heels of Iceland, getting ever closer to "eliminating" Down syndrome in their population.[15]

In the U.S., a 2012 review of the literature on this topic, looking only at U.S. data, found a range from 61% up to 93% of those diagnosed with Down syndrome in the womb who were aborted.[16] More recent data show that abortion accounts for a 33% reduction in the numbers of babies with Down syndrome born in 2014. This means that in recent years there were 33% fewer babies with Down syndrome born in the U.S. than could have been.[17]

Similar rates of discriminatory abortion and attitudes for selection against life are seen for babies diagnosed in the womb with other genetic conditions, as noted before for Trisomies 13 and 18. Janvier and colleagues have documented how medical providers often withhold care for babies prenatally diagnosed with genetic disorders such as Trisomy 18 and Trisomy 13, making an *in utero* "lethal" determination into a reality.[18] Janvier *et al.* also found significant bias among medical professionals toward abortion of unborn children diagnosed with genetic abnormalities. They found that in a survey of delivery room nurses, neonatal intensive care unit nurses, pediatric residents and obstetric residents, 76% would choose termination of pregnancy if the unborn child was diagnosed

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[15] Sarah Zhang "The Last Children of Down Syndrome. Prenatal testing is changing who gets born and who doesn't. This is just the beginning." *The Atlantic* December 2020; accessed at: https://www.theatlantic.com/magazine/archive/2020/12/the-last-children-of-down-syndrome/616928/

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with Trisomy 18, 56% for trisomy 21, and 37% for Turner syndrome.[19] Indeed, a study in *Critical Care Medicine* noted that what doctors tell parents about their child's prognosis is often influenced by the doctor's own attitude toward neurological impairment.[20] There is also evidence that as prenatal testing becomes more prevalent, parents begin to use it to screen subsequent pregnancies for the presence of genetic disease; this has been seen even with monogenic disorders such as Cystic Fibrosis.[21,22]

The problem of relying on genetic screening tests rather than true diagnostic tests is compounded by the fact that the screening tests have significant error rates; one widely-utilized NIPT screening test on the market has a positive predictive value (PPV) of 81%, meaning that there is a significant chance that a positive test result is NOT a true positive.[23] But even this reported PPV value is deceiving, because PPV is based on test sensitivity, specificity, *and* the prevalence of the condition in the population being tested. Because the prevalence of Down syndrome increases with maternal age, PPVs will be higher in patients of advanced maternal age (>35 years old) and will likely increase when other aneuploidy risk factors are known (*e.g.*, ultrasound abnormalities).[24]

A comprehensive study across 21 different U.S. centers of 1,914 women (mean age 29.6 years) observed much lower positive predictive values of 45.5% for trisomy 21. This indicates that a significant proportion (over 50%) of "positive" test results for Down syndrome may not be truly positive when screening women mostly at low risk.[25] For this reason, the authors of this study highlight the "need for follow-up diagnostic testing to confirm true positive results before decisions are made about irrevocable clinical intervention." They know that a woman might tragically abort her child based on an erroneous and incorrect NIPT lab result.

As prenatal genetic tests become more available and expand on the genetic conditions that can be detected, there will undoubtedly be increased pressure to abort unborn children with detected genetic abnormalities. It is easy to imagine that genetic blood disorders such as thalassemia or sickle cell anemia could be targeted, neurological conditions such as Tay-Sachs or Krabbe syndrome, sex chromosome disorders such as Turner and Klinefelter syndrome, Noonan syndrome which affects facial characteristics and the heart, and Osteogenesis Imperfecta (brittle bone disease.)

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Yet these prenatal diagnoses consider the unborn child's worth based simply on her genetic condition, and often are ill-informed of the facts. Contrast the prevalent medical attitude about Down syndrome that leads to a lethal diagnosis, with the reality of recent facts about increased life span, health, learning and functional abilities, including people with Down syndrome who enjoy lifelong work until retirement from their job.[26] Moreover, a study by Skotko *et al.* found that when it came to satisfaction for those with Down syndrome and their families, 99% of people with Down syndrome are happy with their lives, 99% of parents said they love their child with Down syndrome, and 97% of brothers/sisters, ages 9-11, said they love their sibling.[27]

Moreover, while older texts say that around 90% of children born with Trisomy 18 don't live as long as a year, this is simply outdated information. For example, Bella Santorum, daughter of former U.S. Sen. Rick Santorum, turned 12 last May 2020; the title of one story noted that doctors had said her condition was "incompatible with life."[28] Indeed, more and more children with genetic conditions like Bella are surviving, and thriving.[29] A paper by doctors at the Children's Hospital of Philadelphia, published in the journal *Pediatrics,* points out the improvements, noting: "Despite the conventional understanding of these syndromes as lethal, a substantial number of children are living longer than 1 year and undergoing medical and surgical procedures as part of their treatment."[30]

Medical science has also improved significantly in terms of potential interventions, through neonatal and fetal surgeries, potential pharmaceutical treatments as well as cell-based and genetic therapies.[31] We need to consider these young individuals as valued human lives. Eliminating young lives is not the answer to eliminating disease and disability.[32] Destroying the patient is not curative medicine. As Dr. Diana Bianchi, a pioneer in prenatal genetic testing, has noted, we need to "develop new approaches to fetal treatment."[33]

SB 1457 would provide necessary, distinct protections for developing human beings, preventing discrimination based on genetics or disability. Thank you for the opportunity to contribute to the discussion on this important issue.

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