S.247 Testimony to House Commerce & Economic Development Committee
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Good morning Chair Marcotte and members of the House Commerce and Economic Development Committee. My name is Debra Leonard. I am here today to ask for your support of S.247, a bill that would protect Vermonters from discriminatory practices based on their genetic information.

To introduce myself, I am the Chair and Professor of the Department of Pathology and Laboratory Medicine at the University of Vermont Health Network and the Robert Larner College of Medicine at the University of Vermont. In addition to my medical training, I have a doctorate in molecular biology, which is the study of genetic material used to control cell and body functions. My medical specialty is called Molecular Pathology or Genomic Medicine, which focuses on testing of genetic material from patients for medical purposes, including for cancer, infectious diseases and genetic diseases.

I would like to take a bit of time to provide some basic information about genetics and genetic testing. Genetic information is written in an alphabet with just four letters, called A, C, G and T for short. Each person has a genome consisting of about 3 billion of these letters that contains the information for making and running a human body. Every cell in someone's body contains all 3 billion letters of their genome. The parts of the human genome that act as a blueprint for making proteins that are building blocks of the body and do much of the work in the body are called genes. Humans have about 20,000 genes in their genome. Each gene is present in two copies per genome. Each person's genome has variations that makes each of us different from anyone else, unless you have an identical twin. Those genomic variations create unique nose shapes, skin, hair and eye colors, and can create small or large dysfunctions in someone's body. A single letter change (called a single nucleotide polymorphism or SNP, pronounced snip) may do nothing noticeable, or can lead to a disease, such as Sickle Cell Disease. Other diseases are caused by more complex changes in a person's genomic letters. Dominant genetic diseases occur when only one of the two gene copies is changed and the person gets the genetic disease. Recessive genetic diseases occur only if both gene copies are changed; therefore, someone can be a "carrier" of one genetic change and will not get the disease. If their one gene change is passed to their child, the child could also be a "carrier" with only one changed gene copy, or if the second gene copy from the other parent also has a change, then the child will be at risk for the genetic disease. Inherited genetic changes may determine that someone will get a genetic disease, but most often only increases the chance or risk of a genetic disease. The level of certainty that a genetic change will cause a disease is called the "penetrance" of that genetic change. The increased disease risk is in comparison to the general population's risk of getting the same disease when they do not have such a genomic change. When letter differences are present in all the genome copies in a person's body they are called germline changes.

New genomic changes also can occur in a single cell of a person's body that cause that cell and all the cells derived from that cell to either grow faster or live longer than the original cells, thus causing a cancer. The genomic changes that cause cancer can be inherited resulting in an increased risk of certain types of cancer, like changes in the BRCA genes that cause an increased risk of breast cancer. Or the cancer causing changes canbe related to environmental factors such as asbestos exposure

causing increased risk of mesothelioma or smoking causing increased risk of lung cancer. This bill would only relate to testing of a person's germline genome, not cancer genome testing.

What is a genetic test? Many types of genetic test are now used. A genetic test may look for a single letter change or SNP, some or all changes in a single gene, some or all changes in a set of genes, all genes (called the exome), or the entire genome sequence. Evaluation of different amounts of the genome are needed for different disease risks and for different purposes of the testing.

Why do genetic testing? If someone has symptoms of a genetic disease, genetic testing is used to confirm the diagnosis. Identifying the specific genetic change causing the symptoms is useful for testing of other family members to determine if they too have the mutation and an increased riak of disease. Knowing the specific genetic change also may tell healthcare providers something about the course of a disease, the severity of the disease, or even treatment options that may be effective. For example, for a child with repeated trouble breathing and respiratory infections, genetic testing may confirm a diagnosis of Cystic Fibrosis. If the specific mutation is a G552D change, then treatment with a drug called Ivacaftor can be used. This type of genetic testing is called diagnostic testing. In this case, the person has manifested the disease, and the disease diagnosis would be in the medical record of the patient and could be used for life insurance underwriting, like all diagnoses in a person's medical record.

Sometimes someone has family members who have a genetic disease, and they want to know if they are at risk of also getting the disease. In the absence of symptoms, genetic testing can be complex depending on whether the family member who already has been diagnosed with the disease had genetic testing that identified the genomic change related to the disease in their family. This is needed because many genetic diseases can be caused by several or many different genes. Unless the family's genetic change is known, a negative test will not be able to differentiate whether the familial change is not present, or the testing did not include the gene with the familial change. **This type of testing is called predictive genetic testing, is done prior to disease symptoms and usually is NOT deterministic of getting the disease except in rare cases. Even if someone has the genetic change, they have not manifested symptoms of the disease, and may never develop the disease.** The knowledge of having the genetic change that has caused disease in family members allows for monitoring for early onset of the disease, and interventions to prevent or treat the earliest stages of the disease when outcomes are usually better. All these steps would only prolong the life of someone with a genetic disease, which would benefit insurance companies.

Why test people for genetic diseases when they do not have symptoms or a family history of a genetic disease? This is called genomic population screening, as we are doing at the University of Vermont Health Network. Genetic diseases are generally rare and underdiagnosed. Some people do not know the diseases in their blood relatives, such as individuals who are adopted, or when egg or sperm donors were used for conception. Other families are small, and since genetic risks are generally not deterministic, the disease may not have developed in the few family members with the genetic change. This occurs with breast cancer due to BRCA gene changes, when women are the only child with no or few aunts and uncles and no occurrences of breast cancer in their family.

How is genetic testing different from a blood test, such as a cholesterol level test? The usual blood testing, or testing of other body fluids like urine, are only a point in time result that pertains only to that patient at that time. Cholesterol levels can go up or down depending on diet, treatment, and yes, even genetics. In contrast, a genetic test result will never change, since your germline genome is constant. In addition, your genome is shared with your blood relatives, which is not true for a routine blood test such as cholesterol levels. I would argue as a physician, with backing from the American Medical Association and the Vermont Medical Society, that this information critical to someone's health and healthcare deserves additional protections from misuse that could not only affect the individual, who cannot control or change their genome, but also their family members.

When I joined the University of Vermont Health Network in 2013, I came with a bold vision to incorporate genetic information into routine patient care, to identify disease risks, allowing for disease prevention through lifestyle or treatment options, monitoring for disease onset, and treatment at earlier stages of a disease that generally results in better outcomes. This vision aligned with Vermont's goals at the time of assuring greater fairness and equity in how we pay for health care, and improving the health of Vermont's population, moved to action in Act 48 of 2011.

On November 1st of 2019, we began offering genomic screening testing to patients through our primary care providers, working to integrate genetically-determined health risks into the care of our patients. While each individual genetic disease may be rare and affect few people, when taken together, genetic diseases as a group are not rare. To date, we are identifying dominant disease risks in about 20% of the Vermont population, and recessive disease carriers for about 80% of Vermonters. Dominant disease risks identified include cancer, cardiomyopathy, heart rhythm diseases, and muscle diseases. Recessive carrier diseases include spinal muscular atrophy, iron overload, and cystic fibrosis. These results are being used for the healthcare of these individuals, to reduce the impact of these diseases on Vermonters.

Is adverse selection a significant concern? Life insurance representatives in previous hearings on this bill have raised the concern that prohibiting the use of genetic information in underwriting would create adverse selection and financial harm to insurance companies. Genetic information is rarely deterministic of disease or disease severity, but life insurance companies do not focus on disease, just risk of death. Genetic information even more rarely predicts early death of an individual. Only 1.5-2% of the US population is estimated to have a genetic disease that would cause early adult mortality. Of these individuals, many will already know (as will their life insurance company) of their potentially lethal genetic disease risk because of their family history – seeing their parents or grandparents die of the disease. Therefore, a new discovery of a lethal genetic disease from genetic testing would be even rarer than 1.5-2% of the population. Adverse selection is likely to be a rare event for insurance companies. Yet, the impact of early knowledge of all the other genetic disease risks that do not cause early death and may be prevented or treated can be of great benefit to Vermonters and the Vermont health care providers, and, quite frankly, to the insurers who will make more money from patients who live longer, healthier lives.

A Federal Law, the Genetic Information Nondiscrimination Act (GINA) of 2008, protects Americans from health insurance discrimination and employment discrimination based on their genetic

information, but does not protect against other forms of genetic discrimination. Today, we are required to warn people of these remaining risks of discrimination based on their genetic information through informed consent. Because of a lack of full discrimination protections, many people choose not to have genetic testing, despite the potential importance of this information for their health and the health of their families. This fear and declining of genetic testing has been clearly demonstrated and is not just a supposition, as is the fear of adverse selection stated by life insurance companies.

A systematic review of research on genetic discrimination and life insurance (33 studies over 20 years) found the following: 48% of studies concluded that genetic discrimination has some empirical basis, its incidence is rare and it is not a significant source of denials; 42% of the studies documented clear evidence of genetic discrimination in life insurance leading to serious concern; and 9% found no evidence of genetic discrimination in life insurance. Most importantly, the review clearly demonstrated that public fear of genetic discrimination in life insurance underwriting is real. Whether you believe this fear is founded in fact or not, this fear prevents people from agreeing to have genetic testing. This is the medically significant issue. People may choose not to have a genetic test that could benefit their health and healthcare, and that of their family members, because of fear of genetic discrimination in life insurance, and other types of insurance. Both the American Medical Association and the Vermont Medical Society (letters provided to this committee) support genetic discrimination protections for patients for life insurance, long-term care insurance and disability insurance.

I also would like to address statements and recommendations made by the Department of Financial Regulation (DFR). The testimony states, "While S. 247 would allow access to test information that has resulted in a diagnosis, it denies access to test information that may be predictive of significant risk but has not yet resulted in a diagnosis." The United Kingdom (UK) has struggled with this same issue of use of genetic information for insurance underwriting, since the UK uses genetic information in healthcare through their National Health Service. In the UK, insurers cannot require the disclosure of predictive test results unless the specific conditions are pre-approved by the government after consultation with independent experts. The UK only allows requirement of genetic test results for conditions that are caused by changes in a single gene and have an adult onset and high penetrance. Currently, Huntington Disease is the only disease that meets these criteria. This demonstrates the rarity of the importance of genetic diseases in insurance underwriting. Furthermore, the DFR suggests a change in the bill not allowing a request or requirement regarding genetic test information for insurance underwriting. Insurance companies still have access to the medical record, which could reveal results of a predictive genetic test, without a disease diagnosis. Therefore, this suggestion does not provide full genetic discrimination protections to Vermonters. The potential for "hiding" predictive genetic test results outside of the medical record, as was done in the HIV era when I trained in New York City, is a very dangerous practice that can lead to suboptimal healthcare due to a lack of access to all of a patient's medical information. From a medical perspective, this is not a sound approach for protecting Vermonters.

We strongly urge the Committee members to weigh the health care benefits for a majority of Vermonters against the minimal and hypothetical financial risks to the insurance industry, and decide to protect Vermonters from the fear and risk of genetic discrimination, as has already been done in other laws in Vermont. This is the heart of the policy question for lawmakers – do we protect the insurance companies from the VERY SMALL potential risk of adverse selection, or do we protect the health of Vermonters by ensuring confidence in genetic testing that can be used in healthcare to monitor for, prevent or better treat serious genetic diseases?

S.247, if passed, would more fully protect Vermonters from discrimination based on their genetic information when accessing life, long-term care and disability insurance, and help ensure Vermonters feel safe having genetic testing to inform their health and health care. As we are already moving forward with broader use of preventive genetic testing here in Vermont, these protections will be important for Vermonters to benefit from this advancement in health care.

Thank you for your consideration of this very important legislation.

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