

Good morning Senator Lyons and members of the Senate Health and Welfare Committee. My name is Leah Burke. I am here today to ask you for support for S.197, introduced by Senator Ingram, a bill that would help protect Vermonters from discriminatory practices based on their genetic information.

I am a Clinical Geneticist and for the past 20 years have been the head of the Clinical Genetics Program at what is now the University of Vermont Health Network. I am also a Professor in the departments of Pediatrics and Medicine at the Larner College of Medicine at the University of Vermont, where I have taught genetics and genomics to medical students, residents, and fellows in training. Nationally, I chair the Council on Genetics for the American Academy of Pediatrics.

Within the Genetics Program at the University of Vermont Health Network, I examine, diagnose and treat patients with suspected or confirmed genetic conditions. As part of that, I do mostly diagnostic genetic and genomic testing, but the family history is a very important tool in the work that we do and therefore the clinical care extends to that family. Once a genetic diagnosis is made in a patient, there may be a clinical indication to test other family members. For instance, sometimes, if a condition is caused by two genetic changes in a gene, the parents need to be tested to determine whether the changes are on the same copy or on each of the copies of the gene. Additionally, because of the variability in the expression of a genetic condition, a parent may actually have the condition without having the same signs or any signs at all as their child. Having this information is clinically important for the child as well as for the parents for giving accurate recurrent risks. Therefore we often place genetic information in the charts of unaffected individuals. In addition, the family histories that we place in a patient's chart often contain genetic information about their family members. As I mentioned, a family history is considered as important to a patient's care as the past medical history.

My position also involves overseeing and collaborating with genetic counselors. The genetic counselors see individuals with conditions that are thought to be genetic for diagnostic testing. They also see patients and families for counseling around their family histories and possible genetic testing based on those family histories. We provide counseling and arrange testing for the persons coming in for counseling and also members of their families. Most of the time, the individuals seen for genetic counseling and tested because of a family history are adults. There are recommendations against testing minors for adult onset conditions that run in the family, but as in much of life, there are exceptions that need to be made. For example we have had a babies or young children, who are going into the child protective system or being placed up for adoption, who have a known genetic condition in one of their parents. It is in the child's best interest to have as complete a medical history as possible in those situations, so genetic testing for a later onset condition may be done. The child is not symptomatic and may never develop symptoms, but need to have specific monitoring done to look for symptoms that may appear. Without the genetic testing information, the family history may be lost or misinterpreted and the child may have a delay in diagnosis. In particular, one child had a parent with a hereditary cancer. The child was tested and was found to carry the cancer predisposition mutation and therefore will receive earlier screening than they would have with a general population risk.

The patients and families that we see want to know their genetic risk so that they can have proper screening and make life and reproductive decisions. Their family member's genetic testing results are placed in their charts so that the laboratories doing the testing can provide more specific and accurate testing. Even if they decide to wait on genetic testing, the family result is often placed in their chart. They need protection against discrimination for their own pre-symptomatic testing result as well as the results of family members.

We have found that having genetic testing results allows for a more specific diagnosis that in turn allows us to give more accurate prognostic information, and screening practices, and allows the families to participate in clinical trials. Having the ability to test in family members who are not symptomatic assists us in that effort.

Finally, a word about newborn screening programs. Newborn screening programs, Vermont's included, now include conditions that involve genetic testing and may have a later onset. For the traditional newborn screening conditions, such as inborn errors of metabolism, the tests done were primarily biochemical and not genetic, and treatment was initiated right away. With the new conditions, such as spinal muscular atrophy, Pompe disease, and X-linked adrenoleukodystrophy, the testing done up front includes genetic testing and in these conditions, the asymptomatic period can last for years and treatment is not begun right away. So now we have a public health initiative that automatically does genetic testing in the pre-symptomatic phase of disease. A public health initiative that automatically affects all children born in Vermont and may have an adverse effect on their insurability.

You were given a handout containing the informed consent that we have all patients or proxies sign before proceeding with genetic testing. Informed consent is required by both Vermont and New York as well as most of the clinical genetic testing laboratories we use. The last paragraph warns against the discrimination they may encounter as a result of the genetic testing we are recommending. It is my hope that this bill may result in our ability to eliminate or at least alter that paragraph. I applaud your forward thinking in proposing S.197 and expanding the protection against discrimination to include genetic and genomic information. I would be happy to answer any questions.