Robert S. Wildin M.D. for March 30, 2022.

Good afternoon, Chair Lippert and members of the House Health Care Committee. I am grateful for the opportunity to explain today why I ask you to support S.247, a bill to prevent discrimination based on inborn and unchosen genetic characteristics, before they cause symptoms or manifest in a diagnosable disease.

I am a Medical Geneticist. I specialize in the diagnosis, management, and counseling of individuals and families affected by genetic disease. My clinic is at the Children's Specialty Center of the UVM Medical Center in Burlington. I and my geneticist and genetic counselor colleagues see patients of all ages who already have genetic disease, or who have symptoms that might signal a genetic condition. We also see healthy people who have a blood relative diagnosed with a genetic disease and are worried they may come down with the same thing. People who want to expand their family worry about passing something on they may not even know they have. Genetic testing is one of the tools we offer them, but it comes with a catch for those who don't have any symptoms: in seeking the knowable risks, they can lose access to resources and services many of us take for granted, like life insurance. It is difficult to watch as people try to make the best decision for themselves and their families under this odd paradox.

For most of my career, the only way to prevent disease was through family planning and, for a few very rare disorders, special diets. This is changing. More and more treatments exist for both rare and common genetic conditions. At the beginning of my career, children with Cystic Fibrosis rarely lived past 20 and people with Marfan Syndrome often died before 35. Now people with Marfan Syndrome detected early have a normal life span, and thanks to improved care and amazing breakthrough medicines, people with Cystic Fibrosis may live to be older than you or I.

Cancer is more treatable when it is detected at early stages. Early treatment also costs less and has less impact on one's family and livelihood. The same is true for genetically driven disease. Marfan Syndrome and Cystic Fibrosis have early signs that astute health care providers can recognize. But many genetic disorders destroy silently, undetected in routine care. Genetic susceptibility leads to advanced cancer at young ages when it's not on anyone's radar. Susceptibility to heart rhythm disorders can end teenagers' lives suddenly, something that if known in advance, is preventable. Screening for genetic susceptibility, either in those with disease in their family or with DNA screening tests, allows for early medical treatment. Even without preventative options, education for self-efficacy helps people at risk recognize warning signs and, when they occur, seek care immediately.

Sometimes a person with a genetic risk is labeled with a "diagnosis" because it helps their health care providers manage their preventative care. It is important to recognize that a person who has a genetic signature of increased risk should not be considered "diagnosed" for the purposes of this bill if they do not *manifest* a related health condition. For example, someone who inherits a Lynch Syndrome gene variant does not *manifest* that disease until they are found to have a related cancer, which may never happen. A person inheriting a Huntington Disease gene who has sufficient symptoms that would lead to a physician diagnosing them through a physical examination, whether or not they have genetic testing, would have manifested their disease. GINA, the Federal Act prohibiting genetic discrimination for health insurance and employment, describes this distinction well. I understand this was the topic of some discussion when the Committee did their first walkthrough of S.247. It may make sense to emulate GINA, using terms such as "manifest" as opposed to solely relying on "diagnosed" in order to draw a clearer distinction.

We don't know everything about how to use information hidden in one's DNA to improve health and well-being. But we know enough to make a difference, we manage uncertainty, and we learn something every day. People deserve a choice to have that information and share it with their health care providers without fear that it will disadvantage them in their desire to protect themselves and their family.

Fear of negative consequences, real or imagined, has dramatically impacted willingness to take even simple measures that reduce the risk death or disability. We've experienced that intensely over the last year. Within the realm of genetics, scientific studies have found that people often choose not to get genetic testing because legal protections against non-health uses of the information are incomplete. Amazingly, this State officially recognizes this gap as real: both Vermont and New York require people seeking medical genetic testing to sign a written informed consent that includes information on the risk of non-health insurance discrimination. The State also recognizes the value of early detection and treatment: in the routine process called Newborn Screening we screen every baby to prevent death and permanent disability from a small number of treatable genetic disorders.

I am here to support S.247 because it will help people to gather and use their own private genetic information, to take control of their own risks, without fear of losing something else.