Understanding NH Title X – Public Health Chapter 141-H – Genetic testing

Background:

NH Title X – Chapter 141 – H addresses conditions of genetic testing, use of genetic tests in employment situations, use of genetic tests in health insurance, and use of genetic testing in life, disability income, and long-term Care insurance.

Section 141-H:1 sets out definitions of genetic tests. Genetic testing is defined as a test, examination, or analysis which is generally accepted in the scientific and medical communities for the purpose of identifying the presence, absence, or alteration of any gene or chromosome, and any report, interpretation, or evaluation of such a test, examination, or analysis.

Chapter 141-H does not clarify in Section H:1, definitions, or elsewhere if the statute is intended to apply to germline, somatic genetics, infectious agent genetics or all genetic tests. It defers this understanding to the understanding generally accepted in the scientific and medical communities. Germline genetics refers to those genes intrinsic to the whole person and are inherited. Somatic genetics refers to genes that are not passed on but expressed in certain tissues such as tumors. Infectious agent genetics refers to genes intrinsic to infectious pathogens.

It is important to clarify our institutional understanding of the scope of NH Title X – Public Health Chapter 141-H as this impacts our ability to participate in required registries, advance patient care, and perform important research. We believe based on the generally accepted understanding in the scientific and medical communities as well as a logical reading of other sections of NH law genetic testing in Chapter 141-H must only pertain to germline testing and not include somatic or pathogen testing.

Understanding in the scientific and medical community:

The Task Force on Genetic Testing in the United States was commissioned by the National Institutes of Health – Department of Energy Working Group on Ethical, Legal, and Social Implications of Human Genome Research to review genetic testing in the United States and make recommendations to ensure the development of safe and effective genetic tests. The Task Force defined genetic testing is the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites ***in order to detect heritable disease***- related genotypes, mutations, phenotypes or karyotypes for clinical purposes. Such purposes include predicting risk of disease, identifying carriers and establish prenatal and clinical diagnosis or prognosis… ***Tests conducted purely for research are excluded from the definition, as are tests for somatic (as opposed to heritable) mutations, and testing for forensic purposes.***

This definition, used by a key US Task Force, clearly states that genetic testing refers to genes and chromosomes conveying a predisposition to disease (germline genetics), not the genetics of a tumor or infectious agent.

Similarly, the CDC defines genetic testing. The initial definition is not very specific, but clearly indicates that genetic testing requires genetic counseling. Genetic counseling is offered to address concerns related to germline mutations. Testing of tumors does not require genetic counseling unless there are implications for the diagnosis of hereditary conditions.

> "What is Genetic Testing?
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> Genetic testing looks for changes, sometimes called mutations or variants, in your DNA. Genetic testing is useful in many areas of medicine and can change the medical care you or your family member receives. For example, genetic testing can provide a diagnosis for a genetic condition such as Fragile X or information about your risk to develop cancer. There are many different kinds of genetic tests. Genetic tests are done using a blood or spit sample and results are usually ready in a few weeks. Because we share DNA with our family members, if you are found to have a genetic change, your family members may have the same change. *Genetic counseling before and after genetic testing can help make sure that you are the right person in your family to get a genetic test, you're getting the right genetic test, and that you understand your results."*

Again, the understanding of genetic testing by the CDC means testing for mutations or variations in genes that confers a risk or predisposition to disease (germline mutations). It does not include testing the genetics of a tumor (somatic genetic testing).

Here is how the National Cancer Institute defines genetic testing, clearly highlighting inherited changes, and breaking out the concept of tumor DNA sequencing as a separate entity.

> "What is genetic testing?
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> Genetic testing looks for specific inherited changes (variants) in a person's genes. Genetic variants can have harmful, beneficial, neutral (no effect), or unknown or uncertain effects on the risk of developing diseases. Harmful variants in some genes are known to be associated with an increased risk of developing cancer. These inherited variants are thought to contribute to about 5 to 10% of all cancers.
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> Cancer can sometimes appear to "run in families" even if it is not caused by an inherited variant. For example, a shared environment or lifestyle, such as tobacco use, can cause similar cancers to develop among family members. However, certain patterns that are seen in members of a family—such as the types of cancer that develop, other non-cancer conditions that are seen, and the ages at which cancer typically develops—may suggest the presence of an inherited susceptibility to cancer.
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> Genes involved in many of the known inherited cancer susceptibility syndromes have been identified. Testing whether someone carries a harmful variant in one of these genes can confirm whether a condition is, indeed, the result of an inherited syndrome. Genetic testing is also done to determine whether family

members who have not (yet) developed a cancer have inherited the same variant as a family member who is known to carry a harmful (cancer susceptibility predisposing) variant.

A different type of genetic testing, called tumor DNA sequencing, is sometimes done to determine if cancer cells of people who have already gotten a cancer diagnosis have genetic changes that can be used to guide treatment. Although some of these cancer cell changes may be inherited, most occur randomly during a person's lifetime. Genetic testing of tumor cells is addressed in the Biomarker Testing for Cancer Treatment page."

On the NCI Biomarker page, we see:

"What is Biomarker Testing"

Biomarker testing is a way to look for genes, proteins, and other substances (called biomarkers or tumor markers) that can provide information about cancer. Each person's cancer has a unique pattern of biomarkers. Some biomarkers affect how certain cancer treatments work. Biomarker testing may help you and your doctor choose a cancer treatment for you.

There are also other kinds of biomarkers that can help doctors diagnose and monitor cancer during and after treatment. To learn more, visit the Tumor Markers fact sheet.

Biomarker testing is for people who have cancer. People with solid tumors and people with blood cancer can get biomarker testing.

Biomarker testing for cancer treatment may also be called:

tumor testing
tumor genetic testing
genomic testing or genomic profiling
molecular testing or molecular profiling
somatic testing
tumor subtyping

Biomarker testing is different from genetic testing that is used to find out if someone has inherited mutations that make them more likely to get cancer. Inherited mutations are those you are born with. They are passed on to you by your parents."

The last paragraph is critical, in stating that genetic testing is NOT the same as tumor genetic testing. This fits with the Task Force and CDC definitions of genetic testing, specifying that genetic testing refers to inherited conditions and not somatic (tumor) or infectious agent genes.

New Hampshire's Department of Health and Human Services also concurs that "genetic testing" is referring to "the analysis of human DNA, RNA, or chromosomes in order to detect heritable disease-related genotypes, mutations, or karyotypes for clinical purposes." Heritable disease genetics means germline genetics and not tumor or infectious agent genetics.

For internal coherence of Title X:141, genetic testing can only be interpreted as germline genetic testing, and not tumor (somatic) or infectious agent genetic testing.

The understanding of genetic testing throughout Title X must refer only to inherited (germline) conditions and not to tumor genetic testing identifying somatic mutations or infectious agent genetics. A narrow understanding of genetic testing referring only to germline testing for inherited conditions is consistent with the reporting requirements to the NH State Cancer Registry and the surveillance duties of the NH Molecular and Diagnostics Laboratory.

The NH State Cancer Registry is required to collect various tumor markers for NH DHHS, include molecular tumor markers such as HER2, ER, PR, KRAS, JAK-2, Ki-67. These markers reflect somatic (tumor) genetic testing.

The collection of standard variables by the NH State Cancer Registry is required by Title X: Chapter 141-B:7

> "141-B:7 Reporting. – All facilities shall provide a report to the cancer registry, including social security numbers if persons were given the option at the original point of collection to provide social security numbers voluntarily, containing information regarding a cancer diagnosed or being treated. Each report of cancer shall include items listed in rules adopted under RSA 541-A in accordance with the elements listed in the North American Association of Central Cancer Registries, Standards for Cancer Registries as amended from time to time and available."

The list of legally required variables (currently v23) can be found here.

Title X: 141-B:7 (CHRONIC DISEASE PREVENTION, ASSESSMENT AND CONTROL) requires the NH State Cancer Registry to collect, for example, JAK-2 somatic mutation data and requires providers to report that information. This would be in direct conflict with any interpretation of Title X: 141-H (GENETIC TESTING) that might try to extend the definition of genetic testing to include tumor genetic testing.

If the Title X:141-H definition of 'genetic testing' were interpreted as including 'tumor genetic testing' then it follows that Title X: 141-B requires all NH providers diagnosing or treating cancer patients to break the law by reporting that information.

Similar to reporting required by Title X Chapter 141, the NH Molecular Diagnostics Laboratory performs disease surveillance through a variety of tests, including genetic testing, on samples obtained from patients. The NH Molecular and Diagnostics Laboratory surveys a variety of infections including COVID-19, Hepatitis C, and sexually transmitted infectious. Under their public health authority they do not obtain patient consent for testing. NH DHHS requires providers to report, regardless of patient consent, incidences of over fifty diseases, many of which are diagnosed with genetic tests. It follows that Title X

must intend a more narrow definition of genetic testing than, “the presence or absence of a gene or chromosome,” otherwise, the remainder or Title X would be self-contradictory.

Regarding Deidentified Data

US Health and Human services provides guidance for de-identification “in accordance with the Health Insurance Portability and Accountability Act of 1996 (HIPAA) Privacy Rule.” §164.502(d) permits a covered entity to create information that is not individually identifiable. This mitigates risk to individuals and supports the secondary use of data for comparative effectiveness studies, policy assessment, life sciences research, and other endeavors. §164.514(a)-(b) articulates the de-identification standard and implementation specifications.

Use of Genetic Testing in Research

Germline genetics have a stricter standard when being used in research than tumor genetics or infectious agent genetics. Because NH Title X – Chapter 141 – restricts the disclosure of germline testing or its results these data can only be used for research after having been de-identified in accordance with the HIPAA Privacy Rule §164.514 and can no longer reasonably be associated with an individual.

Tumor and infectious agent genetics are also protected as private health information but may be used as other PHI may be used in research. The restrictions of NH Title X – Chapter 141 do not apply to these data.

With written consent the use of genetic data of any type may be used more broadly as allowed by the scope of the consent.

Conclusion:

NH Title X – Chapter 141-H limits the disclosure of genetic testing, particularly as it relates to employment and insurance discrimination. The chapter does not specify if it intends genetic testing to mean all tests related to genes and chromosomes or a subset of test that relate to genes and chromosomes. The chapter’s own definition relies on the scientific and medical communities’ understanding of which tests are included. As detailed above, the scientific and medical communities understand genetic testing to refer to testing related to germline genetics which transmit risks or predispositions for certain diseases. The scientific and medical communities do not understand the term genetic testing to refer to somatic (tumor) genetics or the genetics related to infectious agents. A key NH state health agency (NH DHHS) has published their interpretation of ”genetic testing” as being limited to germline testing.

Additionally, a broader reading of genetic testing would render other requirements of Title X self-contradictory while a more narrow understanding of genetic testing relating only to germline genetics would support an internal coherence to Title X.

Dartmouth Health should interpret Title X, Chapter 141-H as referring to germline genetics only and not include somatic or infectious genetics. Dartmouth Health should develop policies that honor the necessary privacy restrictions of Title X, Chapter 141, relating to germline genetics but not extend restrictions into somatic (tumor) or infectious genetics which are not covered by Title X, Chapter 141.

Germline genetic testing and results have greater restrictions in the setting of research and should only be disclosed in the setting of de-identification in accordance with the HIPAA privacy rule. Tumor (somatic) and infectious agent genetic tests are PHI and may be used in research in the same manner that other PHI may be used. When patient written consent has been obtained genetic data may be used more liberally as per the specific consent.