A WHITE PAPER ON THE NEED FOR **CONSISTENT TERMS FOR TESTING** IN PRECISION MEDICINE

A multi-stakeholder working group of patient advocacy organizations, professional societies, pharmaceutical and diagnostic companies, and laboratories provides recommendations for adoption of consistent, plain language terms for biomarker and germline genetic testing that are applicable across cancer types to help eliminate patient confusion about testing for precision medicine.

PATIENT ADVOCACY GROUPS

AliveAndKicking · CancerCare (Over 75 Years of Help and Hope) · Cancer Support Community · Cholangiocarcinoma Foundation · The Clearity Foundation · CLL Society · Colorectal Cancer Alliance · FIGHT Colorectal Cancer · FORCE (Facing Our Risk of Cancer Empowered) · ican (international cancer advocacy network) · Leukemia & Lymphoma Society · The Life Raft Group · Lymphoma Research Foundation · Living Beyond Breast Cancer · LungCAN · LUNGevity (Find it. Treat it. Live.) · NLCRT (National Lung Cancer Roundtable) · Pancreatic Cancer Action Network · Prostate Cancer Foundation (Curing Together.) · PMC (Personalized Medicine Coalition) · OCRA (ovarian cancer research alliance) · Sharsheret · Susan G. Komen (Where the end of breast cancer begins.™)

PROFESSIONAL SOCIETIES

ACCC (Association of Community Cancer Centers) · AMP (Association for Molecular Pathology) · ASCP STRONGERTOGETHER · National Society of Genetic Counselors · IASLC (International Association for the Study of Lung Cancer — Conquering Thoracic Cancers Worldwide)

INDUSTRY PARTNERS

abbvie · AMGEN · AstraZeneca · Bayer · blueprint MEDICINES · Boehringer Ingelheim · Bristol Myers Squibb · CARIS Life Sciences · Daiichi-Sankyo · Lilly · Foundation Medicine · Genentech (A Member of the Roche Group) · gsk (do more feel better live longer) · Integrated Oncology (LabCorp Specialty Testing Group) · myriad (When Decisions Matter) · Novartis · NeoGenomics · Pfizer Oncology · PGDx (Personal Genome Diagnostics) · ThermoFisher Scientific

With thanks to WS Collaborative for drafting the Working Group White Paper

WS Collaborative — Connecting today's health nonprofits with industry and government

Numerous leaders from the oncology field, including oncologists, nurses, surgeons, pathologists, experts in communications and health disparities, and patient advocates **have endorsed the recommendations in this white paper.**

> Communication is everything; and words matter. Nowhere is clear communication more important than with the diagnosis of cancer and its repercussions; and at no time has clarity been so vital, as we now have targeted molecular therapies for many cancers—therapies that can add months or even years of life. This working group of experienced and engaged stakeholders has made crucial first steps in leveling the playing field for cancer patients and their families to help remove the confusion surrounding biomarker testing and molecular therapy, so patients are educated, energized, and empowered to quickly pursue appropriate testing and subsequent personalized cancer treatment.
>
> TIMOTHY CRAIG ALLEN, MD, JD, FCAP, PROFESSOR AND CHAIR, DEPARTMENT OF PATHOLOGY, THE UNIVERSITY OF MISSISSIPPI MEDICAL CENTER

> Bringing such a diverse set of stakeholders together is quite a feat in itself, yet guiding the difficult work to come to consensus and acceptance of a set of shared terminology in this complex area of cancer care is an incredible accomplishment. Now, in service to patients, the use of these consistent terms (in concert with clear explanations) is foundational to helping them navigate these challenging issues and supporting them in their own information seeking and decision making.
>
> LINDA FLEISHER, PHD, MPH, ASSOCIATE RESEARCH PROFESSOR, HEALTH COMMUNICATIONS AND HEALTH DISPARITIES, FOX CHASE CANCER CENTER

> The massive paradigm shift in cancer care towards precision and targeted therapies includes rapidly evolving use of biomarkers and biomarker testing across the cancer care continuum. The oncology nursing profession supports the essential need for consistent and standardized terminology with easily understood terms for patients and families as they navigate the maze of precision oncology. Nurses spend more time with patients and families than any other member of the health care team and must adopt a common language and avoid jargon and inappropriate and confusing terminology.
>
> PATRICIA FRIEND PHD, APRN-CNS, AOCNS, AGN-BC, ASSOCIATE PROFESSOR AND PROGRAM DIRECTOR, MARCELLA NIEHOFF SCHOOL OF NURSING, LOYOLA UNIVERSITY CHICAGO

> Engaging a patient advocacy community for Lynch syndrome hereditary cancer mutations, we have seen first hand the importance of consistent testing terminology. Our patient community requires a wide variety of genetic and genomic testing and care by a number of different clinical specialists. The first step in navigating the complex care they need is to make sure that all stakeholders are consistent in their understanding of that care. We applaud the diverse working group collaboration in coming together to address this very important issue.
>
> ROBIN DUBIN, EXECUTIVE DIRECTOR, ALIVEANDKICKN, A LYNCH SYNDROME HEREDITARY CANCER PATIENT ADVOCACY ORGANIZATION

Full potential of targeted therapies in lung cancer can only be realized when all patients undergo molecular biomarker testing. This simplistic white paper, developed with widespread collaboration will markedly increase awareness of biomarker testing amongst the oncology cadre, lung cancer caregivers and patients, and the community at large. This is one of the essential steps in the complex process of optimal care of patients with lung cancer with oncogenic drivers.

CHANDRA BELANI, MD, PROFESSOR OF MEDICINE AND ONCOLOGY AT PENN STATE COLLEGE OF MEDICINE AND PENN STATE CANCER INSTITUTE AND CHIEF SCIENCE OFFICER, INTERNATIONAL ASSOCIATION FOR THE STUDY OF LUNG CANCER (IASLC)

Far too many patients across all cancer types are still missing out on essential tests for biomarkers and inherited mutations indicating cancer risk. With rates of biomarker testing and genetic testing for an inherited mutation at sub-optimal levels for numerous patient populations, patients are not benefiting from biomarker-directed care or not learning about their inherited cancer risk. Confusion around testing terms is a driving factor in this undertesting and ultimately has a detrimental impact on patient care.

MICHELLE SHILLER, DO, AP/CP, MGP, CO-MEDICAL DIRECTOR OF GENETICS AT BAYLOR SAMMONS CANCER CENTER AND STAFF PATHOLOGIST AT BAYLOR UNIVERSITY MEDICAL CENTER.

As we search for the ultimate goal of 'personalized medicine' we have begun to understand how the a patient's own genes as well as the molecular features of their cancer can be used to predict risks, disease behavior and response to treatment. Clinicians and patients need to understand how this new information can be helpful in selecting the best strategies in diagnosis, treatment and surveillance. This project makes a significant contribution in simplifying and explaining some of the complex terminology in this field, and will help enhance our understanding of this rapidly evolving field.

SEAN CLEARY, M.D., CONSULTANT, ASSOCIATE PROFESSOR OF SURGERY, CHAIR, DIVISION OF HEPATOBILIARY & PANCREAS SURGERY, VICE CHAIR EDUCATION, DEPARTMENT OF SURGERY, PROGRAM DIRECTOR - HEPATOBILIARY AND PANCREATIC SURGERY FELLOWSHIP, MAYO CLINIC

The treatment of cancer has evolved such that there is no one treatment plan that is best for all patients. Not all pancreas cancers are the same, not all bile duct cancers are the same, not all lung cancers are the same, etc. etc. Thus, it is imperative that we learn the specific genetic make-up of each patient's tumor so that we can personalize the treatment plan to obtain the best possible outcome for our patients. Given the importance of testing for biomarkers and other germline mutations, it's absolutely critical that providers are using consistent terms that patients and their caregivers will start to recognize and understand in order to support a strong foundation for the importance of testing in the patient's care.

SHISHIR KUMAR MAITHEL, MD, PROFESSOR OF SURGERY, DIVISION OF SURGICAL ONCOLOGY, DEPARTMENT OF SURGERY, EMORY UNIVERSITY SCHOOL OF MEDICINE AND SCIENTIFIC DIRECTOR, EMORY LIVER AND PANCREAS CENTER

Introduction

The analysis of a cancer patient's biospecimen (e.g. solid tissue, body fluid, and/or blood) to evaluate for specific driver mutations, multiple gene alterations, and/or non-genomic biomarkers, has made broad application of precision medicine possible in the fight against cancer. However, many eligible patients are not benefiting from biomarker-directed care due to suboptimal testing practices, caused in part by confusion about testing purpose, types and timing of results relative to the start of therapy. This confusion, partially fueled by the disparate testing terminology landscape, is pervasive.[1] Addressing this challenge and creating an action plan have been identified as priorities by leading patient advocacy organizations from across the spectrum of cancer types.

Research has shown that the disparity among terms used to describe testing is one of the patient-identified reasons contributing to confusion and lack of engagement among patients to communicate with providers about testing, leading to less than optimal management of cancer.[2] Developing consistent terminology can reduce patient confusion, improve communication, facilitate shared decision making, support value-based care and assure concordance in policy development.

In pursuit of these objectives, LUNGevity Foundation engaged a variety of stakeholders specializing in various cancer types in a working group to evaluate the current terminology landscape, identify the multitude of terms in use, and leverage their expertise and input from patients to recommend consistent, plain language terms for testing characteristics of a malignancy. This includes testing for somatic (acquired) mutations, proteins, functional tests, genomic signatures, and other biomarkers[3] and testing for germline (inherited) mutations[4]. The working group included leaders from 20 patient advocacy groups representing solid/hematologic malignancies, three professional societies, and 18 pharmaceutical and diagnostics companies and laboratories.[5]

In developing its recommendations, the working group identified 33 terms[6] related to biomarker, genetic and genomic testing being used in patient education and clinical care within the different cancer communities and across stakeholders. Variations in terminology are complicated by the variety of testing modalities, source of samples, overlapping terminology, and the multiplicity of gene mutations that can currently be identified by testing.

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[1] Ferris, A., Mantel, S., Jacobsen, M., Basu Roy, U. "Need for consistent language around biomarker testing in the diagnosis and treatment of lung cancer." International Association for the Study of Lung Cancer World Lung Poster, (September 2016). https://www.lungevity.org/sites/default/files/file-uploads/testing-terminology-world-lung-2016-poster.pdf

[2] *Ibid*

[3] Examples of other biomarkers in cancer include PD1/PDL1 (abnormal protein expression, not necessarily genetic in nature); epigenetic alterations; TMB/MSI/HRD (signatures that are genomic in origin but converted to a "composite score")

[4] Somatic (NCI definition): An alteration in DNA that occurs after conception. Somatic mutations can occur in any of the cells of the body except the germ cells (sperm and egg) and therefore are not passed on to children. These alterations can (but do not always) cause cancer or other diseases. Germline (NCI definition): A gene change in a body's reproductive cell (egg or sperm) that becomes incorporated into the DNA of every cell in the body of the offspring. Germline mutations are passed on from parents to offspring. Also called germline variant.

[5] Appendix I – Working group members.

[6] Appendix II – List of terms that the working group evaluated per landscape or framework assessment results in Appendix III.

Ultimately, working group members agreed on two umbrella descriptor terms. **"Biomarker testing"** was selected as the preferred term for tests that identify characteristics, targetable findings or other test results originating from malignant tissue. **"Genetic testing for an inherited mutation"** and **"genetic testing for inherited cancer risk"** were selected as consensus terms for tests to identify germline mutations (sometimes referred to as variants).[7]

This paper reviews and summarizes the working group's efforts, providing support for its recommendations and a plan for dissemination and implementation of its conclusions. Target audiences for this paper include:

- Patient advocacy groups
- Providers[8]
- Clinical practice organizations
- Industry
- Policymakers
- Payers

Call to Action

Laboratory testing to learn key characteristics about tumors and hematologic malignancies and an individual's risk for hereditary cancers has become a cornerstone of precision medicine in oncology care. Results from these tests can direct treatment decisions (including which treatments may be more relevant and effective based on the patient's tumor characteristics), satisfy enrollment criteria for clinical trials of promising novel agents, and help individuals and families understand and manage their inherited risk for certain cancers.

Guidelines for detection and treatment of cancers across tumor types regularly include recommendations (aimed at providers and patients) encouraging appropriate testing, while cancer patient advocacy organizations have prioritized efforts to expand awareness of and access to testing for their constituents. Expanding timely and appropriate use of testing is a critical component of strategies for reducing death and suffering from cancer and supporting value-based care.

Despite the widespread acceptance within the provider community of the importance of testing, actual testing rates lag far behind best practice recommendations. There are multiple reasons for under-utilization of testing for biomarkers that can direct cancer treatment and risk identification[9]. In our 2019 landscape assessment, we identified 33 terms currently in use to communicate with patients about testing for germline mutations, somatic mutations, and other biomarkers. In many cases, multiple terms

[7] While the genetics community often uses the more technical term "variants" in this context, the working group opted to use "mutation" given its research on what term would be most effective in communicating with patients.
Richards, S. Aziz, N. et al. "Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology." Genetics in Medicine (May 2015).

[8] Types of providers could include Oncologists, Nurses, Nurse Navigators, Genetic Counselors, Surgeons, Pathologists, Molecular Pathologists, Pharmacists, and many other specialist physicians that diagnose or treat patients with cancer.

[9] Mason, C. Ellis, P.G. et. al. "Patterns of Biomarker Testing Rates and Appropriate Use of Targeted Therapy in the First-Line, Metastatic Non-Small Cell Lung Cancer Treatment Setting" J Clin Pathw. 2018 Jan-Feb; 4(1): 49–54.

were used to describe the same test. With so many terms in use, it is not surprising that patients and caregivers are confused about what kind of testing to ask for, what kind of testing they may have had, whether they have received the appropriate testing for their specific condition, and what this testing means for their care plans. Common examples include patients confusing germline genetic and genomic testing or not understanding the need for repeat testing for somatic mutations and other biomarkers after a failed line of therapy or disease progression.

To best serve the needs of cancer patients who are extremely vulnerable and often overwhelmed by their diagnoses and treatment decisions, it is time to harmonize language, simplify communications and clearly explain the goals of testing. It is incumbent on all parties involved to work together to use clear and consistent terminology, from the testing manufacturers and laboratories who originate the tests, the pharmaceutical industry that develops and markets related therapies, providers who care for patients, guidelines agencies that promulgate best practice recommendations, payers that communicate about coverage and make payment decisions, policy makers who create regulatory and coverage guidelines, and the patient advocacy community that serves patients directly.

A unified voice and message will help the medical community and patients achieve common understanding about the use and potential impact of testing to drive care decisions while increasing patient empowerment and satisfaction, a generally recognized indicator of high-quality care. Working group members are committed to adopting the umbrella terms “biomarker testing” for tests that identify disease characteristics and “genetic testing for an inherited mutation” or “genetic testing for inherited cancer risk” for tests to identify germline (inherited) mutations (sometimes referred to a variants), providing additional tumor or constituency-specific information as needed. We urge all stakeholders to join us in this commitment.

Adopted July 2020, Consistent Testing Terminology Working Group

Background

Testing for acquired mutations and other solid tumor and hematologic malignancy characteristics, also commonly referred to as somatic testing, is key for treatment decision-making in many types of cancer – particularly for those with FDA-approved biomarker-directed standard of care therapies. Some examples include PDL-1-based immunotherapy, BRAF inhibitors, PARP inhibitors, EGFR inhibitors, and numerous other targeted therapies, to name only a few. For multiple tumor types, the importance of laboratory testing, particularly comprehensive multi-biomarker testing, at diagnosis of advanced disease and at disease progression/recurrence, is underscored by recent therapeutic advances that include tumor-type agnostic therapies approved for patients with relatively rare genomic mutations (e.g., larotrectinib for TRK fusion-driven cancers and pembrolizumab for microsatellite instability (MSI) or MSI-high cancers). Such comprehensive testing can also enable identification of patients eligible for clinical trials because many drugs are being evaluated in patients whose tumors have specific characteristics.

Despite the imperative for testing to support the ability of patients and providers to make informed decisions about treatment options, too many patients across cancer types are still not receiving testing at diagnosis, after progression/recurrence or as part of the treatment decision-making process.

For example, although there are FDA-approved therapies for multiple specific sub-types of **lung cancer**, recent data indicates that only 7% of patients receiving care in community oncology practices/programs, where the vast majority of cancer patients are treated, received comprehensive testing for all seven of the biomarkers recommended in the NCCN guidelines at the time of publication.[10] There is a low frequency of single-gene biomarker testing in **gastrointestinal stromal tumors (GIST)** patients with only 26.7% tested for the KIT mutation as recommended by the National Comprehensive Cancer Network (NCCN) Guidelines.[11] Additional testing to identify other driver mutations occurs in only 30% of GIST cases that should be considered for testing.[12] Meanwhile, a recent survey of **cholangiocarcinoma** patients found that more than half are not being offered testing,[13] even though there are multiple actionable mutations and therapy-development programs aimed at those mutations within both tumor types. Similarly, recent data indicate that 40% of metastatic **colorectal cancer** patients are not receiving recommended testing.[14]

In addition, while NCCN and other professional societies publish guidelines for genetic testing for inherited cancer risk, testing rates remain below 50% for most people in those populations covered by current NCCN guidelines, which include people diagnosed with breast cancer at age 45 or younger, triple-negative breast cancer at age 60 or younger, and ovarian, pancreatic, metastatic prostate, or male breast cancer at any age.[15]

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[10] Gierman HJ, Goldfarb S, Labrador M, et al. Genomic testing and treatment landscape in patients with advanced non-small cell lung cancer (aNSCLC) using real-world data from community oncology practices. J Clin Oncol. 2019;37(suppl; abstr 1585).
[11] Florindez J, Trent J. Low Frequency of Mutation Testing in the United States: An Analysis of 3866 GIST Patients. Am J Clin Oncol. 2020 Apr.
[12] Rothschild S, Call J, Corless C, Gill A, Miettinen M, Rubin B. Diagnostic algorithm for gastrointestinal stromal tumor (GIST) using patient registry data impacts pathology guidelines. J Clin Oncol 38: 2020 (suppl; abstr e23519).
[13] Cholangiocarcinoma Foundation. https://www.surveymonkey.com/results/SM-X7ZB2TPBV/
[14] Gutierrez ME, Price KS, Lanman RB, et al. Genomic Profiling for KRAS, NRAS, BRAF, Microsatellite Instability (MSI) and Mismatch Repair Deficiency (dMMR) among Patients with Metastatic Colon Cancer. JCO Precision Oncol. December 2019.
[15] Allison W. Kurian, Kevin C. Ward, Nadia Howlader, et al. Genetic Testing and Results in a Population-Based Cohort of Breast Cancer Patients and Ovarian Cancer Patients. Journal of Clinical Oncology. May 20, 2019.

There are multiple probable reasons for the under-utilization of biomarker testing[16] and genetic testing for inherited cancer risk across cancer types, including limited sample availability, poor processes for biospecimen collection, handling and processing preceding molecular analysis (also referred to as pre-analytics[17]), lack of support for or knowledge about testing among providers, limited access to medical genetics physicians and genetics counselors, complex non-uniform insurance preauthorization policies, lack of decentralized testing and limitations on patients' access to testing stemming from cost barriers and poor insurance coverage. Recognizing the multi-faceted nature of this problem, the working group limited its focus to one specific topic: confusion and lack of understanding among patients exacerbated by a multiplicity of terms used in communicating about testing.

The working group's effort was complicated by multiple challenges, including:

- Some cancer types do not have established NCCN (or other similar) guidelines indicating which biomarker testing should be done.
- Some disease states may be due to either somatic (acquired) or germline (inherited) causes or both.
- In some cancer settings the term "genetic testing" is often used to refer to germline (inherited) mutations, while in other settings the term may be used to refer to testing for somatic (acquired) mutations.
- Some organizations speak to more than one disease state constituency.
- Some groups do not yet have agreement within their organizations on what the best terms should be.

Nonetheless, the group identified clear areas of shared commitment, including:

- Aligning the vocabulary patients, providers, and industry use when referring to testing for germline mutations, somatic mutations, and other biomarkers.
- Increasing patient literacy and reducing patient confusion about laboratory testing.
- Helping patients understand if they have had testing and learn about the value of biomarker testing and genetic testing for inherited cancer risk for their care.
- Empowering patients to ask for the appropriate laboratory testing for their disease state
- Increasing patient (particularly late-stage cancer patient) understanding that some biomarker testing is appropriate at diagnosis, and for some cancer patients, at progression or recurrence.
- Helping to harmonize practice between academic and community institutions.
- Developing collaborative educational materials across cancer types.

[16] The International Association for the Study of Lung Cancer (IASLC) Global Survey on Molecular Testing in Lung Cancer. Smeltzer MP, Wynes MW, Lantuejoul S, et al. J Thorac Oncol. 2020 May 14:S1556-0864(20)30383-X. doi: 10.1016/j.jtho.2020.05.002.

[17] Preanalytics is defined as the collection, handling and processing of clinical specimens obtained during routine procedures in the practice of medicine, surgery, interventional radiology, and pathology to aid in diagnosis, determine treatment choice and monitor therapy. https://www.archivesofpathology.org/doi/full/10.5858/arpa.2019-0009-SA https://link.springer.com/article/10.1007/s40139-018-0179-5

Patient Quotes:

"When people are diagnosed, they don't know any of these terms. . . we need to find answers and we need to find them quick. It makes it difficult that there is not standard terminology." (LUNGevity)

"Being diagnosed with cancer is like being drop-kicked into a foreign country; you don't know where you are, you aren't familiar with the territory or culture and you certainly don't speak the language, yet you need to figure out how to survive! We need simple, consistent language that we can understand and digest to help ease our anxieties and allow us and our loved ones to be engaged and make informed decision about our care." (LUNGevity)

"(Testing terminology is) a matter of semantics and what it 'is' vs. what it's 'for'. The professional community cares about what it is and what it's looking for. The patient community cares about what good it might do them..." (Cholangiocarcinoma Foundation)

"I wish there was a dictionary for all common tumor mutations that was accessible to patients both in terms of getting it and in terms of understanding it." (FORCE)

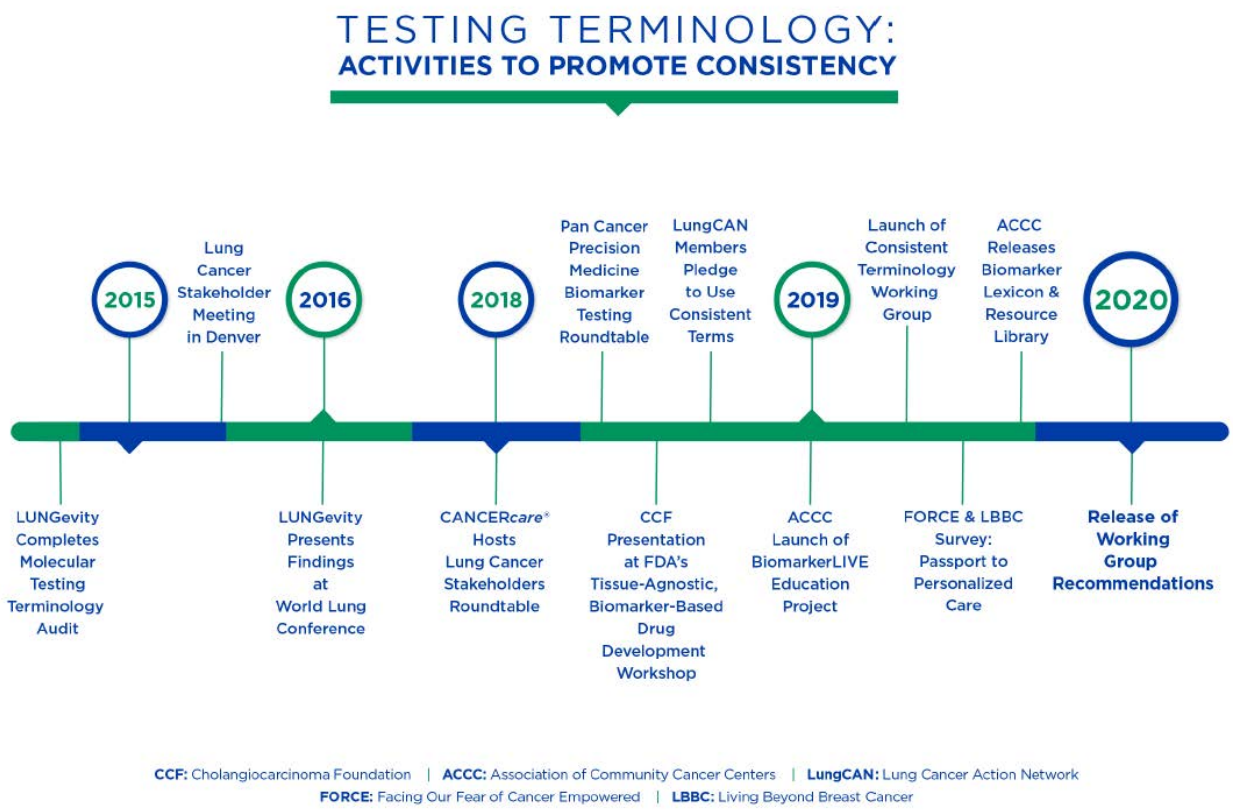
"Some people talk about the TUMOR having a mutation vs. the PATIENT having a mutation. I still don't get it." (FORCE)

"I don't understand the jargon." (FORCE)

Working Group Process

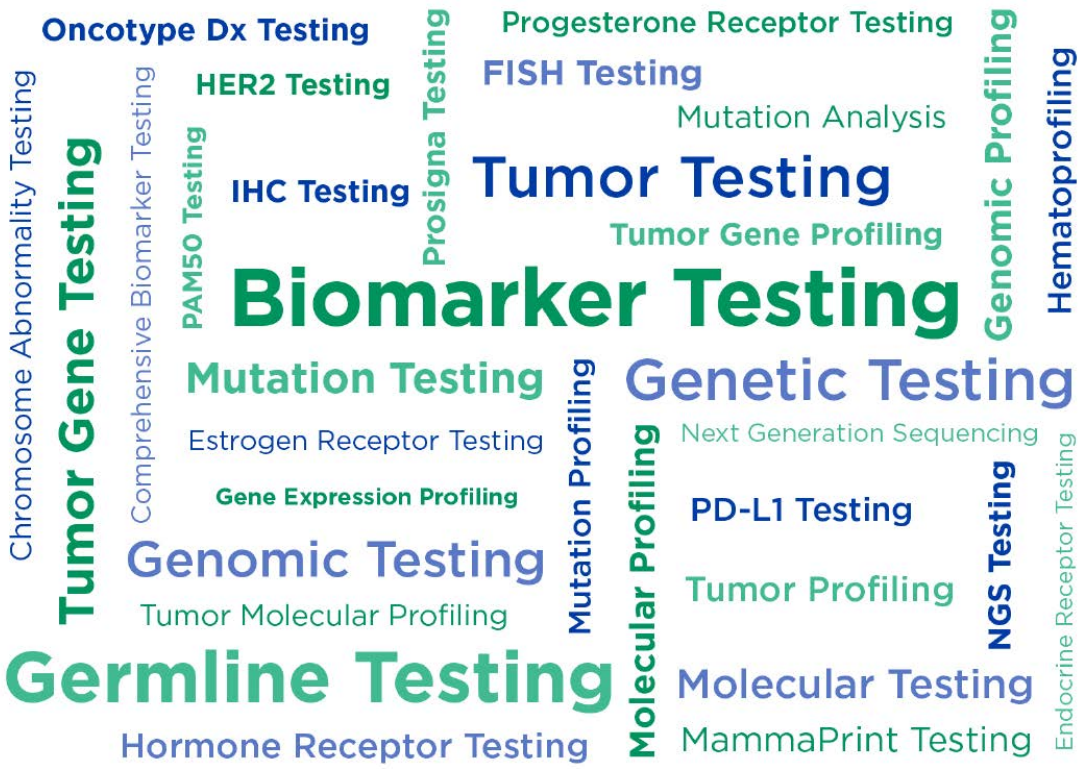
While the working group itself was first convened in April 2019 as an outgrowth of the LUNGevity-convened Pan Tumor Precision Medicine & Biomarker Testing Roundtable (March 8, 2019)[18], multiple activities led by individual patient advocacy organizations and professional societies undertaken over many years formed the groundwork for the working group's effort.

[18] Appendix IV – List of participants at the Pan Tumor Precision Medicine & Biomarker Testing Roundtable and the meeting agenda.



A key milestone culminating from these efforts was the identification of 33 terms currently in use across the cancer landscape to communicate about testing for germline (inherited) mutations, somatic (acquired) mutations and other biomarkers. The list of these terms was compiled through a framework assessment that evaluated:

- Types of tests being used for various cancer types (solid tumors and blood cancers).
- Stages when guidelines recommend the testing be used.
- Type of biospecimen (solid tissue, body fluid, or blood) required to perform testing.
- Purposes for those tests.
- Terms used to describe or discuss the testing.
- Preferred terms adopted by the working group member or applied broadly across each specific disease state's cancer community.



The framework assessment was then leveraged to identify a short list of terms for prioritization by the working group members. (Appendix III) The group met regularly throughout 2019 and early 2020 to discuss pros and cons of the various terms and develop agreed-upon umbrella terms. In parallel, working group members pursued internal alignment within their respective organizations and among their core constituencies to coalesce support for the recommended terms.

Ultimately, the working group agreed to conduct separate discussions on delineating specific terms to be used in describing testing for tumor characteristics, including for acquired mutations and in describing germline genetic testing for inherited mutations and hereditary risk.

Working Group Recommendation 1: "Biomarker Testing" (preferred term for testing for somatic (acquired) mutations and other biomarkers)

In arriving at the recommendation to use the umbrella term "biomarker testing", the group agreed upon a definition focused on testing specimen that originates from the neoplastic tissue. "Biomarker testing" means the laboratory analysis of a patient's biospecimen (solid tissue, body fluid, and/or blood) to test for specific biologically relevant mutations, multiple gene alterations, proteins and/or other biomarkers. Testing can include, but is not limited to, single tests, panel tests, and multi-plex panel tests (such as Next Generation Sequencing, NGS) as supported by medical and scientific evidence.

In selecting "biomarker testing", group members felt that it had the broadest applicability to all types of cancer (solid/liquid cancers) and diverse testing modalities (proteomic, single analyte testing, DNA/RNA sequencing, staining patterns on pathology slides, as well as emerging technologies and methods for

assessing genetic signatures such as tumor mutational burden). In addition, based on the working group's comprehensive framework analysis, "biomarker testing" was the most common term already in use for patient education across patient advocacy groups, professional societies, and industry.

The group considered a variety of other terms that are frequently used to describe laboratory testing designed to identify relevant biomarkers. Among the terms considered was "tumor profiling." While acknowledging the common use of this term, the working group concluded that it is not sufficiently broad to address biomarker testing in blood cancers. In addition, there could be concerns about using the term "profiling" in patients from underserved communities, including racial and ethnic minority groups, where "profiling" has a negative connotation.

The working group also considered the term "molecular testing." However, members concluded this term is too diffuse, as it does not fully encompass all testing approaches. [19] The group wanted to ensure that the umbrella term would be inclusive of all types of laboratory testing used across cancer types, while acknowledging that an individual group could add detail to the umbrella term where relevant (e.g. "biomarker testing including molecular profiling for [specific] biomarkers").

The possibility of adding the modifier "comprehensive" to the umbrella term "biomarker testing" to emphasize the importance of multi-target testing (especially for cancer types such as lung cancer that have multiple targeted therapy treatment options approved and in development) was also evaluated. The group concluded that doing so would add unnecessary complexity and would not be appropriate for all disease states. Rather, individual organizations can promote the umbrella term with additional descriptive detail relevant to their specific disease state. For example, groups could communicate about "biomarker testing including broad panel or NGS testing for [specific] biomarkers."

There were some limitations noted for the term "biomarker testing," specifically that there may be certain disease states such as ovarian and pancreatic cancer where the term "biomarker" may include laboratory testing for monitoring of disease recurrence using laboratory results and computational methods. Two examples are CA125, a protein used in disease monitoring in most ovarian cancer patients, and CEA for monitoring colorectal cancer. For patients in these disease states, using the term "biomarker testing" to address testing for tumor characteristics may result in gaps in patient-provider communication about the patient's testing needs. For these disease states, the group recommends using "biomarker testing" as an umbrella term to introduce the concept of testing for biomarkers, and later explaining the need for the patient to request specific tests, such as "biomarker testing including tumor testing" or "biomarker testing for treatment decisions."

Noting that it may take some organizations and constituencies time to reach alignment, the working group concluded that "biomarker testing" provides the most comprehensive term, broadly applicable to all cancer types (solid and liquid tumors), all testing modalities (including proteomic, DNA and RNA sequencing with next-generation or other technologies), and all medical applications.[20]

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[19] Molecular testing has been taken to mean nucleic acid testing, but could also be used to apply to proteomic or IHC assays, and many clinical chemists will rightly claim that sodium and potassium are molecules. It also leaves out important analyses such as karyotype and does not reflect the active role this testing often plays in clinical decision making.

[20] Biomarker testing can be thought of covering all the relevant categories of lab testing utility:
-Screening (e.g. Lynch syndrome, cervical cancer screening)

Working Group Recommendation 2: "Genetic testing for an inherited mutation" OR "Genetic testing for inherited cancer risk" (preferred terms for germline genetic testing)

The process to arrive at a recommendation for umbrella terms to communicate about germline genetic testing for cancer risk involved multiple steps, given the additional complexities surrounding this topic. Working group members agreed up front about the need to have separate terms in this area, acknowledging that confusion about germline genetic testing is exacerbated by a lack of understanding of the distinction between inherited (germline genetic) mutations and tumor-specific alterations (somatic or acquired mutations) and other biomarkers.

Twenty-five organizations in the working group that specialize in supporting people with inherited cancers and/or in some aspect of conducting germline genetic testing were surveyed on this topic.[21] Based on the survey results, which indicated there may not be consensus for a single umbrella term, the group focused on identifying two terms that together would be acceptable to the working group members.

As the group considered several options for the consensus umbrella term, they identified limitations with the terms "genetic" and "germline" for patient education. The term "genetic" on its own was considered too broad, given that its definition includes any kind of testing that identified chromosome or gene changes. The term "germline," while scientifically accurate, was considered problematic given that most lay people are unfamiliar with its meaning and may be put off by the potential relation to "germs."

Constituent Survey

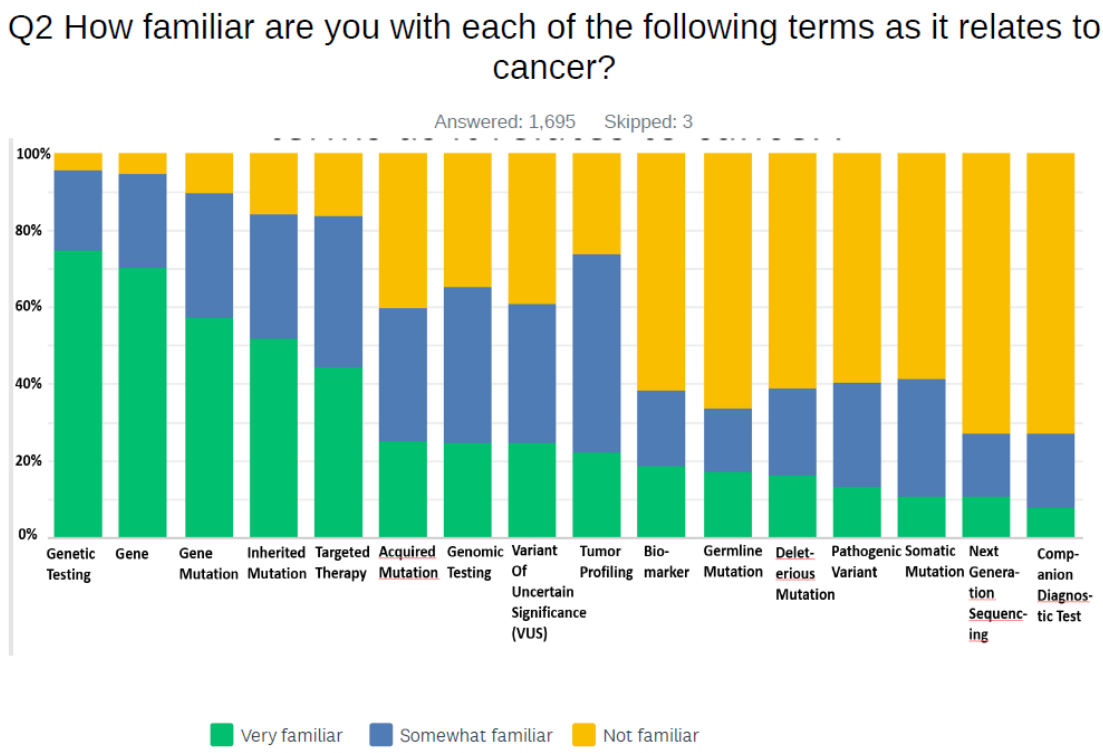
To evaluate what could work best, working group members partnered with Facing Our Risk of Cancer Empowered (FORCE) to survey patients within their respective constituencies. This survey initially generated more than 300 responses within the hereditary cancer community and ultimately yielded almost 1700 responses from within the broader cancer community during the early months of 2020.[22]

The final analysis of the survey data pointed to significant gaps in understanding of the multiple terms commonly used to describe germline genetic testing.

-Diagnostic (e.g. identification of specific fusions in some sarcomas)
-Prognostic
-Predictive (prediction of therapy response)

[21] Appendix VI - Survey results from working group members on germline testing terms.

[22] Appendix VII – Survey results from patients on germline testing terms.



From the survey, two terms emerged as those preferred by the lay cancer community: “genetic testing for inherited cancer risk” and “genetic testing for an inherited mutation.”

In the final analysis of survey respondents, multiple themes emerged that provided helpful insight to the working group. For example, when asked if they had one preferred term, survey respondents wrote in “genetic testing for inherited cancer risk” more often than any other term. The second most common write-in term was “genetic testing for an inherited mutation. More people stated that they objected to the term “germline” than any other term.

Some respondents emphasized the benefit of including “risk” to emphasize that testing positive does not mean a person currently has or will be diagnosed with cancer in the future. People who have not been diagnosed with cancer more strongly opposed the term “genetic testing for hereditary cancer” than people who had been diagnosed with cancer. People who had genetic testing were more likely to oppose the term “germline genetic testing” than people who had not had testing.

Next Steps

Guidelines for detection and treatment of cancers across tumor types often include recommendations (aimed at providers and patients) encouraging testing, and cancer patient advocacy organizations have prioritized efforts to expand awareness of and access to testing for their constituents. Expanding timely use of testing is a critical component of strategies for reducing death and suffering from cancer and enhancing value-based care.

Given the need to expand the use of appropriate testing for cancer patients and those at risk for cancer, the working group's recommendations described in this paper are offered to minimize patient confusion with regard to the testing they should have, the testing they have had, and what the results of their testing may mean for their care decisions.

Working group members hope this work will support a unified voice and message about testing that can help the medical community and patients achieve common understanding.

As next steps, working group members have committed to adopting the umbrella terms "biomarker testing" and "genetic testing for an inherited mutation" or "genetic testing for inherited cancer risk" within our own communications, providing additional tumor or constituency-specific information as needed.

The working group has launched a multi-faceted dissemination and communications effort to ensure its recommendations and supporting materials are widely available among all key stakeholders within the cancer ecosystem, including providers, patient advocacy organizations, guidelines agencies, industry, payers, and policymakers.

This work on consistent nomenclature is a significant step forward in ensuring that all appropriate patients receive biomarker testing and genetic testing for an inherited mutation, and there is still much work to do to bridge the gaps and barriers to testing for all. There is a need for awareness and education on: 1) obtaining adequate and high quality samples for testing, whether that be tissue, cells, or blood, 2) standardizing procedures and protocols for collecting and processing specimens, 3) laboratory quality assurance/control programs, 4) understanding the technical aspects of the various tests and platforms, 5) enhancing communication on testing between members of the multidisciplinary care team, 6) increasing clinician adherence to guidelines for biomarker and germline genetic testing; and 7) interpreting and understanding the testing results relative to providers, patients, and caregivers.

Finally, the working group is considering evolving into a more formal Alliance, led by a multi-stakeholder steering committee. The mission of this alliance would be to jointly address access to optimal care driven by awareness of and access to biomarker testing and genetic testing for an inherited mutation across cancer types and constituencies.

Appendices

Appendix I

Consistent Testing Terminology Working Group Participants

Patient Advocacy Group	Representative
AliveAndKickn	Robin Beth Dubin, Executive Director
American Cancer Society	Lauren Rosenthal, Director, National Lung Cancer Roundtable
Cancer*Care*	Christine Verini, Chief Operating Officer
Cancer Support Community	Claire Saxton, Vice President, Education
The Cholangiocarcinoma Foundation	Stacie Lindsey, President
Clearity Foundation	Deborah Zajchowski, PhD, Scientific Director
Colorectal Cancer Alliance	Andrea Goodman, Vice President, Patient & Family Support Ronit Yarden, PhD, MHSA, Former Senior Director, Medical Affairs
Fight CRC	Reese Garcia, Research Advocacy Manager
FORCE (Facing Our Risk of Cancer Empowered)	Sue Friedman, DVM, Executive Director, Founder Lisa Schlager, Vice President, Public Policy
International Cancer Advocacy Network	Marcia Horn, JD, President and CEO
Leukemia & Lymphoma Society	Beth Davison, Clinical Trial Nurse Navigator, Clinical Trial Support Center
The Life Raft Group	Denisse Montoya, Director, Patient Registry Sara Rothschild, Vice President, Program Services
Lymphoma Research Foundation	Victor Gonzalez, Senior Manager of Support Services
Living Beyond Breast Cancer	Janine Guglielmino, Vice President, Mission Delivery
Lung Cancer Action Network (LungCan)	Dusty Donaldson, Executive Director
LUNGevity Foundation	Nikki Martin, MA, Director, Precision Medicine Initiatives Kristen Santiago, Senior Director, Public Policy Initiatives
PanCAN	Cassadie Moravek, Associate Director, Clinical Initiatives Jenny Isaacson, Vice President, Strategic Partnerships and Projects
Personalized Medicine Coalition	Cynthia Bens, Senior VP, Public Policy
Prostate Cancer Foundation	Andrea Miyahira, Ph.D., Director of Research Becky Campbell, Coordinator Rebecca Levine, Chief of Staff and VP of Government Affairs
Ovarian Cancer Research Alliance (OCRA)	Chad Ramsey, VP, Public Policy Vanessa Cramer, Public Policy

Sharsheret (The Jewish Breast & Ovarian Cancer Community)	Peggy Cottrell, LCGC, Genetic Counselor
Susan G. Komen	Erica Kuhn, MPH, Manager, Education Publications Susan Brown, MS, RN, Sr. Director, Education and Patient Support
Professional Society	**Representative**
Association of Community Cancer Centers (ACCC)	Janelle Schrag, MPH, Senior Program Manager, Provider Education
Association for Molecular Pathology (AMP)	Sarah Thibault-Sennett, PhD, Policy Fellow, Public Policy and Advocacy Eric Konnick, MD, MS, FCAP, Pathologist, Seattle Cancer Care Alliance, UW Medical Center
National Society of Genetic Counselors (NSGC)	Meghan E. Carey, CAE, Executive Director Leila Jamal, ScM, Certified Genetic Counselor, NIAID, Affiliated Scholar, NIH Department of Bioethics Christie Jett, MS, LCGC, Genetic Counselor, Valley Health Rachel Shapira, ScM, LCGC, Genetic Counselor, University of California at Los Angeles (UCLA)
International Association for the Study of Lung Cancer (IASLC)	Becky Bunn, Senior Advisor, Scientific Affairs Murry Wynes, PhD, Senior Advisor, Scientific Affairs
Industry Partner	**Representative**
Abbvie	Amanda Leiting, Associate Director, Companion Diagnostics, Oncology
Amgen	Francesca Angeletti, Director, Global Advocacy Relations
AstraZeneca	Kerri Culton, Director, Oncology Nurse Educators Sara Green, Senior Director, US Advocacy and Alliance, Oncology Julie Ramage, Director, Precision Medicine Quality Initiatives and Partnerships Michelle Cosgrove, Associate Director, US Advocacy and Alliance, Oncology
Blueprint Medicines	Dave Dubinski, Advocacy Relations Elissa Quinn, Precision Medicine
Boehringer Ingelheim	Lara Crissey, Director, Patient Advocacy, Specialty Care Meredith Liberto, Associate Director, Patient Advocacy and Professional Relations, Oncology
Bristol-Myers Squibb	Kemi Osundina, Manager, Advocacy & Policy Michael Cantrell, PhD, Medical Scientist, Biomarkers and Diagnostics Sam Simmons, MD, Regional Director - East Region, Pathology Diagnostic Liaison Team Brenda Yuan, PharmD, Biomarkers & Diagnostics Consultant
Caris Life Sciences	Mark Daras, VP, US Pathology Sales/Solutions
Eli Lilly and Company	Devon McGoldrick, Oncology Advocacy and Professional Relations
Foundation Medicine	Brian Tomlinson, Director, Patient and Professional Partnerships
Genentech	David Cooling, Senior Manager, Alliance and Advocacy Relations Judy Largen, Biomarker Testing Team
GlaxoSmithKline (GSK)	Jeff Emch, Vice President, Diagnostics Strategy

	Jennifer Faikish, Oncology Diagnostics Strategy
Novartis	Karen Hamel, Director, Advocacy Relations
Myriad Women's Health	Ronit Lebor, MS, CGC, Regional Medical Specialist in Women's Health, Upstate New York
NeoGenomics	Gina Wallar, PhD, Senior Vice President, Clinical Division
Pfizer	Marianne Gandee, Senior Director, Team Lead, Patient and Professional Relations Josh Bergren, Director, Advocacy and Professional Relations
Personal Genome Diagnostics (PGDx)	Maura Kadan, RN, Director, Clinical Education and Outreach
Thermo Fisher Scientific	Amy K. Carroll, Ph.D., Medical Affairs Director, North America, Clinical NGS and Oncology Division, Life Sciences Solutions Jody Courtney McIntyre, Associate Director, Oncology Product Management, Clinical Sequencing Division Mauricio Minotta, Director, Public Relations

Appendix II

List of terms that the working group evaluated as a result of the framework assessment exercise (see Appendix III).

1. Biomarker testing
2. Comprehensive biomarker testing
3. Molecular testing
4. Molecular profiling
5. Mutation testing
6. Mutation analysis
7. Mutation profiling
8. Genomic testing
9. Genomic profiling
10. Tumor testing
11. Tumor profiling
12. Tumor molecular profiling
13. Hematoprofiling
14. Tumor gene profiling
15. Tumor gene testing
16. Hormone receptor testing
17. Estrogen receptor testing
18. Progesterone receptor testing
19. Endocrine receptor testing
20. HER2 testing
21. IHC testing
22. FISH testing
23. PD-L1 testing
24. Oncotype Dx testing
25. MammaPrint testing

26. PAM50 testing
27. Prosigna testing
28. Chromosome abnormality testing
29. Gene expression profiling
30. NGS testing
31. Next Generation Sequencing
32. Genetic testing
33. Germline testing

Appendix III

The working group's framework assessment was completed in September-October 2019 and covered solid tumors and hematologic malignancies. It was used to create a short list of terms for evaluation seen in Appendix II.

Disease State	Sub Type	Stage when patient is tested	Desired Biomarker Testing for Somatic Mutations Performed at Diagnosis	Desired Germline Testing Perfo	Desired Biomarker	Source of Sample	Source of Sample for	Purposes of Test for munity	Terms that describe the testing in columns D & E & F	If available, preferred term in your disease space	Groups that have reviewed/edited this content for their disease space
Lung Cancer	Non Small Cell Lung Cancer	3b/4	1. NGS panel for multiple genes EGFR, ALK, KRAS, ROS1, BRAF, NTRK,MET, RET, HER2, TMB 2. IHC test for PDL1 level	None	1. NGS panel for multiple genes 2. Liquid biopsy (using NGS) for multiple genes	Tumor tissue (primary source)	Tumor tissue or blood for liquid biopsy	Identify treatable driver mutation and establish PDL1 protein expression level	1. Comprehensive biomarker testing 2. Molecular testing / profiling 3. Tumor testing / profiling	Comprehensive biomarker testing	LUNGevity, CancerCare, ICAN
Cholangiocarcinoma	Intrahepatic Perihilar Distal	All stages	1. NGS panel for mutations 2. IHC testing for PDL1 level	none	1. NGS panel for multiple mutations 2. Liquid biopsy (using NGS) for multiple genes		Tumor tissue (primary source) or blood for liquid biopsy	Identify treatable driver mutation and establish PDL1 protein expression level	1. Genomic profiling/molecular profiling	Biomarker Testing or Targeted Therapy Testing or Personalized Therapy Testing	Cholangiocarcinoma Fdn ICAN
Ovarian	Epithelial	3/4	BRCA1/2 (will be guideline); preferred:	NGS panel inc multi-gene biomark	NGS panel inc multi-gene bi	Tumor	Tumor tissue (primary source) or blood for liquid biopsy	Identify treatable driver mutation and establish PDL1 protein expression level; identify potential for immune checkpoint inhibitor therapy	1. Tumor molecular profiling 2. Tumor profiling		Clearity, ICAN
Ovarian		All stages		BRCA1/2 + risk gene panel					Germline or genetic testing		Clearity
Colorectal Cancer		3/4	NGS panel for multiple genes including KRAS, NRAS, BRAF, HER2, NTRK, MMR	MSI-H	1. NGS panel for multiple genes 2. Liquid biopsy (using NGS) for multiple genes & epigenetics markers	Tumor tissue	Tumor tissue or blood for liquid biopsy	Identify biomarkers to detect recurrence/prognosis Identify biomarkers to dictate treatment options. Identify potential for immune checkpoint inhibitor therapy	Biomarker testing, tumor testing, germline/genetic testing Molecular Profiling (ICAN)	Biomarker testing, tumor testing	Colorectal Cancer Alliance, Flight CRC, ICAN
Colorectal Cancer		all stages	IHC for dMMR / NGS panel for MMR genes	MSI-H /TMB	(MSI-H Testing or IHC) if positive, PDL1 levels	Tumor tissue / blood	Tumor tissue or blood for liquid biopsy	Identify biomarkers to dictate treatment options. Identify potential for immune checkpoint inhibitor therapy	Biomarker testing, tumor testing, germline/genetic testing	Biomarker testing, tumor testing, genetic testing	Colorectal Cancer Alliance, Flight CRC
Lymphoma/CLL	NHL, HL, CLL	All stages	NGS panel for multiple genes including BCL2, BCL6, MYC, ALK, TP53, MYD88	None	NGS panel for multiple genes	Bone marrow, blood	Bone marrow, blood	To identify biomarkers that will help determine appropriate treatments	Molecular testing or genomic testing Hematoprofiling (ICAN)		LRF, ICAN - Q from CSC - Should we mention that

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Appendix IV

PAN TUMOR PRECISION MEDICINE AND BIOMARKER TESTING ROUNDTABLE ATTENDEES

Patient Advocacy Group / Society Representatives

Monique Dawkins **Association of Community Cancer Centers (ACCC)** Assistant Director, Education Programs	**Marianne Gandee** **Association of Community Cancer Centers (ACCC)** Director, Development and Strategic Alliances
Tara Burke **Association for Molecular Pathology (AMP)**	**Sarah Thibault-Sennett** **Association for Molecular Pathology (AMP)**

Senior Director, Public Policy and Advocacy	Policy Fellow, Public Policy and Advocacy
Claire Saxton **Cancer Support Community** Senior Director, Education	**Mary Ott** **Cholangiocarcinoma Foundation** Research Advocate
Deborah Zajchowski **Clearity Foundation** Scientific Director	**Patrice Brown** **Colorectal Cancer Alliance** Vice President, Programs
Ronit Yarden **Colorectal Cancer Alliance** Senior Director, Medical Affairs	**Reese Garcia** **Fight CRC** Research Advocacy Manager
Lori Tauber Marcus **Kraft Precision Medicine Accelerator, Harvard Business School** Chair, DTP Initiative	**Denisse Montoya** **Life Raft Group** Director, Patient Registry
Jessica Nowak **Life Raft Group** Director, Outreach and Engagement	**Janine Guglielmino** **Living Beyond Breast Cancer** Vice President, Mission Delivery
Andrew Ciupek **Lung Cancer Alliance** Manager, Clinical Research	**Jenny Isaacson** **PanCAN** Vice President, Strategic Partnerships and Projects
Cassadie Moravek **PanCAN** Associate Director, Clinical Initiatives	

Industry Representatives

Robin Burkhart **AstraZeneca** Marketing Manager	**Lise Hall** **AstraZeneca** Associate Director, Consumer Marketing
Philina Lee **Blueprint Medicines** Vice President, Commercial Strategy and Operations	**Raymond Mankoski** **Blueprint Medicines** Vice President, Medical Affairs
Kathryn Byrne **Boehringer Ingelheim**	**Lara Crissey** **Boehringer Ingelheim**

Patient Advocacy Liaison	Director, Patient Advocacy and Professional Relations
Barbara Moehring **Boehringer Ingelheim** Director, Clinical Development Medical Affairs	**Emily Prince** **Bristol-Myers Squibb** Biomarker Diagnostics, Medical
David Marshak **Foundation Medicine** Manager, Patient Advocacy	**David Cooling** **Genentech** Senior Manager, Government Affairs
Elissa Quinn **Genentech** National Key Account Manager	**Karen Hamel** **Novartis** Director, Patient Advocacy
Jackie Rosenbaum **Bristol-Myers Squibb** Associate Director, Advocacy and Policy	

LUNGevity Staff

Dylan Ashley Grants Assistant	**Meriam Driss** Vice President, Strategic Partnerships
Andrea Ferris President and CEO	**Kayla Haskins** Communications Manager
Lisa Justen Grants and Partnerships Manager	**Nikki Martin** Director, Precision Medicine Initiatives
Kristen Santiago Senior Director, Public Policy Initiatives	**Linda Wenger** Senior Vice President, Marketing and Communications

Appendix V

PAN TUMOR PRECISION MEDICINE AND BIOMARKER TESTING ROUNDTABLE
Bethesda North Marriott Hotel & Conference Center
March 8, 2019

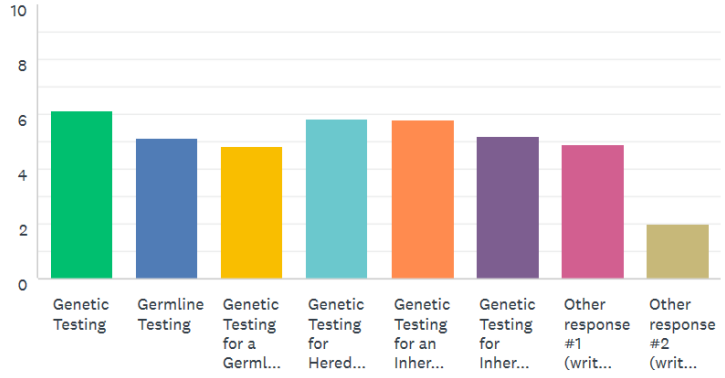
9:30 – 9:40	Welcome and Meeting Overview	Andrea Ferris Nikki Martin
Disease Space Presentations on Biomarker Testing Barriers/Best Practices		
9:40-10:30	Lung Cancer • LUNGevity • Lung Cancer Alliance	Nikki Martin Andrew Ciupek
10:30-10:55	Ovarian Cancer • Clearity	Deborah Zajchowski
10:55-11:05 BREAK		
11:05-11:30	Sarcoma • Life Raft Group	Denisse Montoya
11:30-11:55	Cholangiocarcinoma / Bile Duct Cancer • Cholangiocarcinoma Foundation	Mary Ott
11:55-12:15 WORKING LUNCH		
12:15-12:40	Pancreatic Cancer **(Webinar Presentation)** • Pancreatic Cancer Action Network (PanCAN)	Jenny Isaacson Cassadie Moravek
12:40-1:05	Pan Tumor • Cancer Support Community	Claire Saxton
1:05-1:30	Colorectal Cancer • Fight CRC	Reese Garcia
1:35-1:55	Breast Cancer • Living Beyond Breast Cancer	Janine Guglielmino
1:55-2:20	Professional Society • Association of Molecular Pathology (AMP)	Tara Burke Sarah Thibault-Sennett
2:20-2:30 BREAK		
Discussion on Areas of Commonality		
2:30-3:45	• Which barriers exist across multiple disease spaces • Which challenges are common to multiple groups? • What solutions exist to address barriers?	All
3:45-4:00	Wrap up	Nikki Martin

Appendix VI

Responses to the first germline testing survey on preferred terminology for patient education that 26 working group members who work with patients with inherited cancers completed in October-November 2019. The list of terms that members submitted as alternate options was transcribed directly from Survey Monkey for ease of viewing.

Please rank the terms you prefer to use when educating patients about germline (also commonly referred to as genetic) testing. This term would be the big umbrella term you use when speaking about testing for germline mutations. Please note that you have a chance to rank other terms not listed - just rank the option field and then write in the new term in the field provided in Question #2.

Answered: 25 Skipped: 1



TERMS SURVEYED

- Genetic Testing
- Germline Testing
- Genetic Testing for a Germline Mutation
- Genetic Testing for Hereditary Cancer
- Genetic Testing for an Inherited Mutation
- Germline Testing for Inherited Cancer
- Other Response #1
- Other Response #2

Q2

Save as

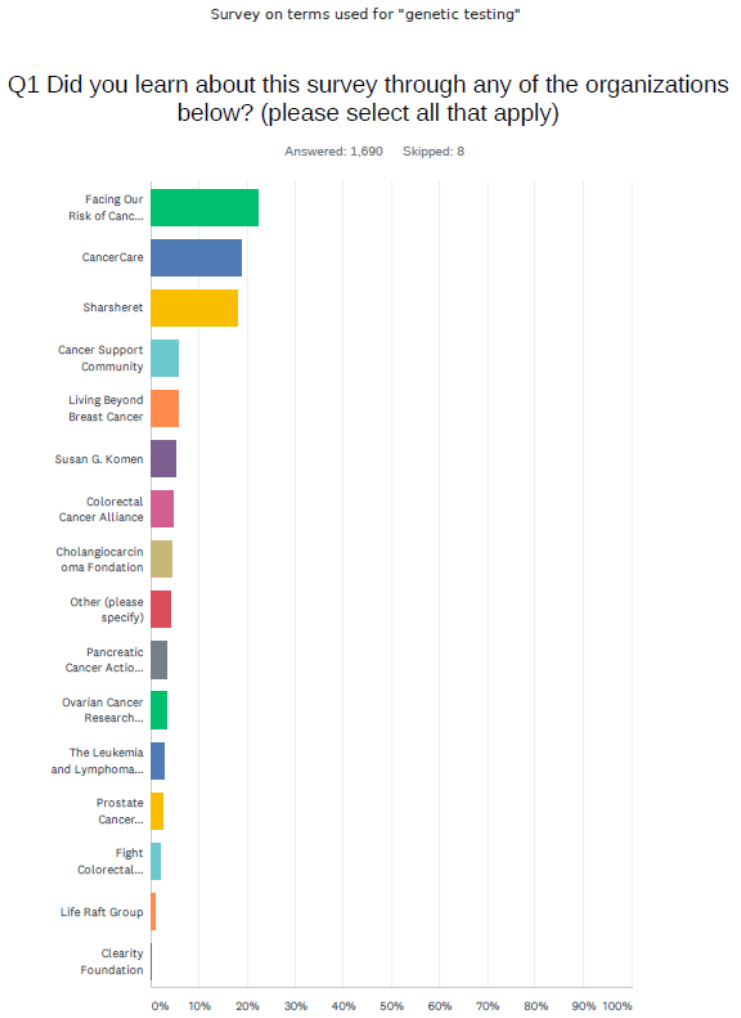
Please ener the "other responses" that you ranked above. Clearly call out your "other #1" and "other #2" if you have more than one new suggestion for terms.

Answered: 17 Skipped: 9

1. N/A
2. NA
3. N/A
4. Germline genetic testing
5. Our educational materials do not currently encompass TAs that reference germline testing
6. No other. Simply “Genetic Testing” is sufficient
7. It's not a patient subtype in focus for us. Most of the breast cancer community uses the qualifier "hereditary" when differentiating between inherited mutations and cancer genomics.
8. Genetic Testing for an Inherited Disease (NOTE: this was how we referred to it in our most recent DTC testing guide which was focused beyond cancer)
9. 1. Hereditary Biomarker Testing (write in option #1) 2. Inherited Biomarker Testing (write in option #2)
10. N/A
11. Reasoning for choosing “genetic testing for inherited mutation” is because germline testing is often done for reasons other than cancer. If we are raking preference for a term that only covers inherited “cancers” then my first preference would be “genetic testing for hereditary cancer”.
12. None
13. Genetic Testing for Cancer You can Inherit [This is 9th grade reading level. Many of the other terms are college reading level. CSC aims for 6-8th grade reading level.]
14. N/A
15. #2 "inherited disorder testing". AMP has members who are involved in germline testing for inherited disorders beyond the cancer-sphere, so we use this term to be inclusive of ALL types of germline testing for inherited conditions/disorders, regardless of if they involve cancer.
16. N/A
17. BRCA 1/2 testing (very specific)

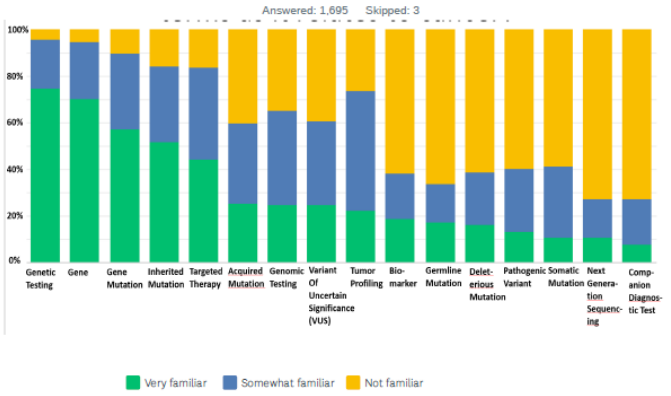
Appendix VII

Results from Patient Germline Testing Survey conducted in January-March 2020



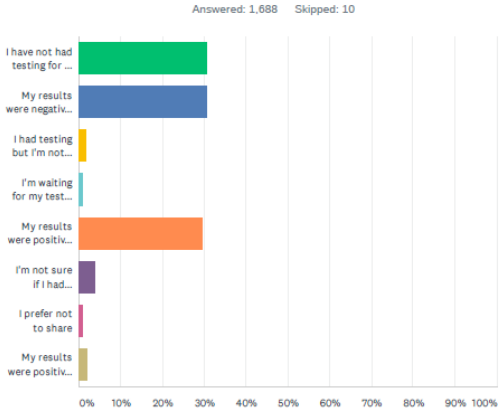
Survey on terms used for "genetic testing"

Q2 How familiar are you with each of the following terms as it relates to cancer?



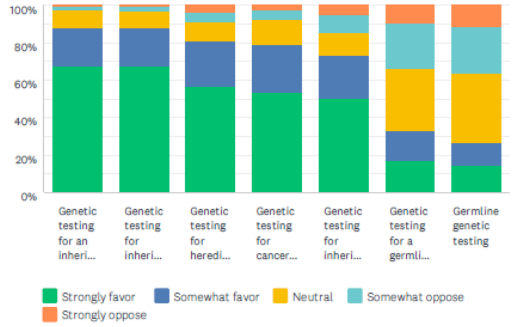
Survey on terms used for "genetic testing"

Q3 Tests can find gene changes (called mutations) that a person may be born with. These changes are called "germline" or "inherited" gene mutations. They can be passed from fathers or mothers to their sons and daughters. Some inherited gene changes increase risk for cancer, and can cause cancer to run in families. Have you had testing for an inherited gene mutation linked to cancer? If so, what were your test results?



Q4 The term "genetic testing" is very general and can be used to describe different types of tests. This can lead to confusion among patients and their health care providers. Adding terms like "germline" or "inherited" may help distinguish tests used to learn if a person has an inherited gene mutation that is linked to cancer risk. If organizations can agree to use one term for the same test, this may reduce confusion. The groups that developed this survey want to help patients communicate with their doctors to get the right tests and treatments for them. In order to find a common term we can use, we are exploring which terms most people find more or less acceptable and clear. Please rate your feelings about the following terms used to describe genetic testing for an inherited mutation on a scale from "strongly favor" to "strongly oppose."

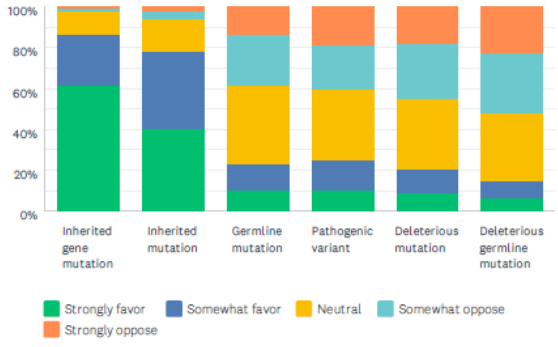
Answered: 1,687 Skipped: 11

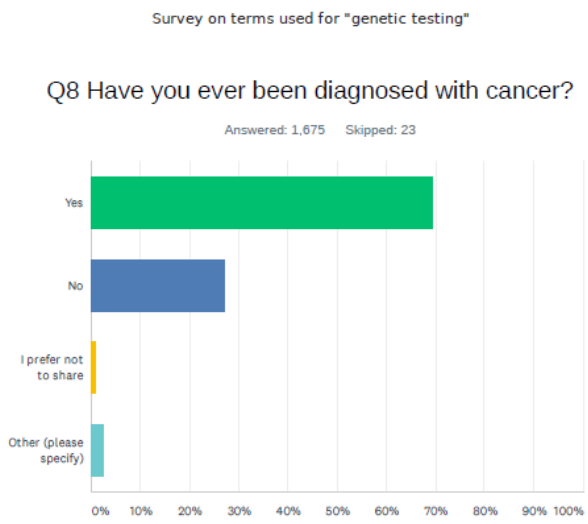


Survey on terms used for "genetic testing"

Q6 Health care professionals and laboratories may also use different terms for inherited gene mutations. The common term "gene mutation" is used broadly, which can lead to confusion. The groups that developed this survey are exploring which terms are more or less acceptable and clear to most people. Please rate your feelings about the following terms used to describe inherited gene mutations on a scale from "strongly favor" to "strongly oppose."

Answered: 1,653 Skipped: 45





Appendix VIII

Consistent Testing Terminology Working Group White Paper Steering Committee

Name	Title	Organization	Email
Beth Davison	Clinical Trial Nurse Navigator, Clinical Trial Support Center	Leukemia & Lymphoma Society	Beth.davison@lls.org
Dave Dubinski	Advocacy Relations	Blueprint Medicines	DDubinski@blueprintmedicines.com
Sue Friedman, DVM	Executive Director, Founder	FORCE - Facing Our Risk of Cancer Empowered	suefriedman@facingourrisk.org
Andrea Goodman, MSW, MPH	Vice President of Patient & Family Support	Colorectal Cancer Alliance	agoodman@ccalliance.org
Stacie Lindsay	President	The Cholangiocarcinoma Foundation	Stacie@cholangiocarcinoma.org
Christie Jett, MS, LCGC	Genetic Counselor	Valley Health	ajett@valleyhealthlink.com
Eric Konnick, MD, MS	Pathologist	Seattle Cancer Care Alliance, UW Medical Center	ekonnick@hotmail.com
Nikki Martin	Director, Precision Medicine Initiatives	LUNGevity Foundation	nmartin@lungevity.org

Julie Ramage	Director, Precision Medicine Quality Initiatives and Partnerships	Astra Zeneca	Julie.ramage@astrazeneca.com
Janelle Schrag, MPH	Senior Program Manager, Provider Education	Association of Community Cancer Centers	jschrag@accc-cancer.org
Ronit Yarden, PhD, MHSA	Consultant, Former Senior Director, Medical Affairs, Colorectal Cancer Alliance		ronityarden@gmail.com