Lexicon for "A.C.T. (Assess, Change, Test) on Molecular Testing"

(Last reviewed June 2016)

Contents

Introduction

The purpose of this lexicon is to define common terms and phrases that appear in the context of molecular biomarker testing in patients with lung cancer. Phrases like "genomic profiling" and "molecular testing" must be clearly understood by members of the cancer care team in order to facilitate clear dialogues and coordinated management plans for patients with lung cancer.

Sources:

Two main sources of information were used for this lexicon:

- The NCI Dictionary of Cancer Terms features 7,850 terms related to cancer and medicine. http://www.cancer.gov/publications/dictionaries/cancer-terms
- American Cancer Society Cancer Glossary: http://www.cancer.org/cancer/cancerglossary/index

Key Terms

ALK (Anaplastic Lymphoma Kinase) Gene

NCI (Cancer.gov)	ACS (Cancer.org)
A gene that makes a protein called anaplastic lymphoma kinase (ALK), which may be involved in cell growth. Mutated (changed) forms of the ALK gene and protein have been found in some types of cancer, including neuroblastoma, non-small cell lung cancer, and anaplastic large cell lymphoma. These changes may increase the growth of cancer cells. Checking for changes in the ALK gene in tumor tissue may help to plan cancer treatment. Also called anaplastic lymphoma kinase gene.[1]	Not found

Alteration

NCI (Cancer.gov)	ACS (Cancer.org)
A change resulting in something that is different from the original.[2]	Not found

Antibody

NCI (Cancer.gov)	ACS (Cancer.org)
A protein made by plasma cells (a type of white blood cell) in response to an antigen (a substance that causes the body to make a specific immune response). Each antibody can bind to only one specific antigen. The purpose of this binding is to help destroy the antigen. Some antibodies destroy antigens directly. Others make it easier for white blood cells to destroy the antigen. An antibody is a type of immunoglobulin.[3]	A protein made by immune system cells and released into the blood to help defend the body against foreign agents, such as bacteria. These agents contain certain substances called antigens. Each antibody works against one certain antigen.

Antigen

NCI (Cancer.gov)	ACS (Cancer.org)
Any substance that causes the body to make an immune response against that substance. Antigens include toxins, chemicals, bacteria, viruses, or other substances that come from	A substance that can cause the body's immune system to respond by making antibodies. For example, the immune system responds to antigens that are part of bacteria and viruses to

outside the body. Body tissues and cells, including cancer cells, also have antigens on them that can cause an immune response. These antigens can also be used as markers in laboratory tests to identify those tissues or cells.[4]	help people resist infections. Certain cancer cells have antigens that can be found by lab tests. Knowledge of these antigens can be used to help diagnose, monitor, and even treat some cancers.

Biomarker

NCI (Cancer.gov)	ACS (Cancer.org)
A biological molecule found in blood, other body fluids, or tissues that is a sign of a normal or abnormal process, or of a condition or disease. A biomarker may be used to see how well the body responds to a treatment for a disease or condition. Also called **molecular marker** and **signature molecule**.[5]	Referenced as "**tumor marker**:" Substances that can be found in the body (in the blood, urine, or other body fluids or tissues) when a person has cancer. These substances are usually made by cancer cells, but are sometimes by normal cells as well. Different types of cancer can have different tumor markers. For example, prostate-specific antigen (PSA) is a tumor marker for prostate cancer. Ideally, a tumor marker would always be found when a person has a certain type of cancer, and would never be found if a person didn't have cancer. But in reality tumor markers are rarely like that, because normal cells can often make them as well.

EGFR (Epidermal Growth Factor Receptor)

NCI (Cancer.gov)	ACS (Cancer.org)
The protein found on the surface of some cells and to which epidermal growth factor binds, causing the cells to divide. It is found at abnormally high levels on the surface of many types of cancer cells, so these cells may divide excessively in the presence of epidermal growth factor. Also called epidermal growth factor receptor, ErbB1, and HER1.[6]	Not found

Gene

NCI (Cancer.gov)	ACS (Cancer.org)
The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein.[7]	A piece of DNA (deoxyribonucleic acid) inside a cell that has the information to make a specific protein. Genes are responsible for traits passed on in families, such as hair color, eye color, and

	height, as well as susceptibility to certain diseases.

Gene Expression Profile

NCI (Cancer.gov)	ACS (Cancer.org)
Information about all messenger RNAs that are made in various cell types. A gene expression profile may be used to find and diagnose a disease or condition and to see how well the body responds to treatment. Gene expression profiles may be used in precision medicine.[8]	Not found

Genetic Testing

NCI (Cancer.gov)	ACS (Cancer.org)
Analyzing DNA to look for a genetic alteration that may indicate an increased risk for developing a specific disease or disorder.[9]	Tests that can be done to see if a person has certain gene changes known to increase the risk of cancer or other diseases. Such testing is not recommended for everyone, but for people with certain types of family history. Genetic counseling should be part of the genetic testing process.

Genetics

NCI (Cancer.gov)	ACS (Cancer.org)
The study of genes and heredity. Heredity is the passing of genetic information and traits (such as eye color and an increased chance of getting a certain disease) from parents to offspring.[10]	Not found

Genome

NCI (Cancer.gov)	ACS (Cancer.org)
The complete set of DNA (genetic material) in an organism. In people, almost every cell in the body contains a complete copy of the genome. The genome contains all of the information needed for a person to develop and grow. Studying the genome may help researchers understand how different types of cancer form and respond to treatment. This may lead to new ways to diagnose, treat, and prevent cancer.[11]	The total DNA (deoxyribonucleic acid) and sometimes RNA (ribonucleic acid) in a single cell, representing all of the genetic information of the organism. The term genome can also refer to certain types of genetic material, such as that in the cell's nucleus (nuclear genome) or mitochondria (mitochondrial genome). See also deoxyribonucleic acid, gene, ribonucleic acid.

Genomic Profiling

NCI (Cancer.gov)	ACS (Cancer.org)
A laboratory method that is used to learn about all the genes in a person or in a specific cell type, and the way those genes interact with each other and with the environment. Genomic profiling may be used to find out why some people get certain diseases while others do not, or why people react in different ways to the same drug. It may also be used to help develop new ways to diagnose, treat, and prevent diseases, such as cancer. **Also called genomic characterization**.[12]	Not found

Genomic Sequencing

NCI (Cancer.gov)	ACS (Cancer.org)
A laboratory method that is used to determine the entire genetic makeup of a specific organism or cell type. This method can be used to find changes in areas of the genome that may be important in the development of specific diseases, such as cancer.[13]	Not found

Genomics

NCI (Cancer.gov)	ACS (Cancer.org)
The study of the complete genetic material, including genes and their functions, of an organism.[14]	Not found

Hereditary

NCI (Cancer.gov)	ACS (Cancer.org)
In medicine, describes the passing of genetic information from parent to child through the genes in sperm and egg cells. Also called inherited.[15]	Not found

Hereditary Mutation

NCI (Cancer.gov)	ACS (Cancer.org)
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. Hereditary mutations are passed on from parents to offspring. Also called **germline mutation.**[16]	Not found

Kinase

NCI (Cancer.gov)	ACS (Cancer.org)
A type of enzyme (a protein that speeds up chemical reactions in the body) that adds chemicals called phosphates to other molecules, such as sugars or proteins. This may cause other molecules in the cell to become either active or inactive. Kinases are a part of many cell processes. Some cancer treatments target certain kinases that are linked to cancer.[17]	Not found

Kinase Inhibitor

NCI (Cancer.gov)	ACS (Cancer.org)
A substance that blocks a type of enzyme called a kinase. Human cells have many different kinases, and they help control important functions, such as cell signaling, metabolism, division, and survival. Certain kinases are more active in some types of cancer cells and blocking them may help keep the cancer cells from growing. Kinase inhibitors may also block the growth of new blood vessels that tumors need to grow. Some kinase inhibitors are used to treat cancer.[18]	Not found

Molecular Marker

NCI (Cancer.gov)	ACS (Cancer.org)
A biological molecule found in blood, other body fluids, or tissues that is a sign of a normal or abnormal process, or of a condition or disease. A molecular marker may be used to see how well the body responds to a treatment for a disease or condition. Also called **biomarker** and **signature molecule**.[19]	Not found

Molecular Test

NCI (Cancer.gov)	ACS (Cancer.org)
In medicine, a laboratory test that checks for certain genes, proteins, or other molecules in a sample of tissue, blood, or other body fluid. Molecular tests also check for certain changes in a gene or chromosome that may cause or affect the chance of developing a specific disease or disorder, such as cancer. A molecular test may be done with other procedures, such as biopsies, to help diagnose some types of cancer. It may also be used to help plan treatment, find out how well treatment is working, or make a prognosis.[20]	Not found

Molecule

NCI (Cancer.gov)	ACS (Cancer.org)
The smallest particle of a substance that has all of the physical and chemical properties of that substance. Molecules are made up of one or more atoms. If they contain more than one atom, the atoms can be the same (an oxygen molecule has two oxygen atoms) or different (a water molecule has two hydrogen atoms and one oxygen atom). Biological molecules, such as proteins and DNA, can be made up of many thousands of atoms.[21]	Not found

Mutation

NCI (Cancer.gov)	ACS (Cancer.org)
Any change in the DNA sequence of a cell. Mutations may be caused by mistakes during cell division, or they may be caused by exposure to DNA-damaging agents in the environment. Mutations can be harmful, beneficial, or have no effect. If they occur in cells that make eggs or sperm, they can be inherited; if mutations occur in other types of cells, they are not inherited. Certain mutations may lead to cancer or other diseases.[22]	A change in the DNA (deoxyribonucleic acid) of a cell. Most mutations do not cause cancer, and a few may even be helpful. But all types of cancer are thought to be due to mutations that damage a cell's DNA. Some cancer-related mutations can be passed on from a parent (inherited). This means that the person is born with the mutated DNA in all the body's cells. But most mutations happen after a person is born. These are called somatic or acquired mutations. This type of mutation happens in one cell at a time, and only

	affects cells that arise from the single mutated cell.
See also: Hereditary Mutation, Somatic Mutation	See also: Somatic Mutation

Next-Generation Sequencing (NGS)

NCI (Cancer.gov)	ACS (Cancer.org)
Not found	Not found
Other sources: • Second-generation sequencing methods that lead to faster, high-throughput sequencing of DNA fragments in unison.[23] • Compared to traditional sequencing, the ability to fully sequence large numbers of genes in a single test and simultaneously detect deletions, insertions, copy number alterations, translocations and exome-wide base substitutions in known cancer-related genes.[24]	

Personalized Medicine

NCI (Cancer.gov)	ACS (Cancer.org)
A form of medicine that uses information about a person's genes, proteins, and environment to prevent, diagnose, and treat disease. In cancer, personalized medicine uses specific information about a person's tumor to help diagnose, plan treatment, find out how well treatment is working, or make a prognosis. Examples of personalized medicine include using targeted therapies to treat specific types of cancer cells, such as HER2-positive breast cancer cells, or using tumor marker testing to help diagnose cancer. Also called **precision medicine**.[25]	Not found

Precision Medicine

NCI (Cancer.gov)	ACS (Cancer.org)
A form of medicine that uses information about a person's genes, proteins, and environment to prevent, diagnose, and treat disease. In cancer, precision medicine uses specific information about a person's tumor to help diagnose, plan treatment, find out how well treatment is working, or make a prognosis. Examples of precision medicine include using targeted therapies to treat specific types of cancer cells,	Not found

such as HER2-positive breast cancer cells, or using tumor marker testing to help diagnose cancer. Also called **personalized medicine**.[26]	

Protein

NCI (Cancer.gov)	ACS (Cancer.org)
A molecule made up of amino acids. Proteins are needed for the body to function properly. They are the basis of body structures, such as skin and hair, and of other substances such as enzymes, cytokines, and antibodies.[27]	A large molecule made up of a chain of smaller units called amino acids. Proteins serve many vital functions inside and outside of the body's cells.

Protein Expression

NCI (Cancer.gov)	ACS (Cancer.org)
Refers to the production of proteins by cells. The study of protein expression in cancer cells may give information about a specific type of cancer, the best treatment to use, and how well a treatment works.[28]	Not found

Somatic Mutation

NCI (Cancer.gov)	ACS (Cancer.org)
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.[29]	A change (mutation) in DNA (deoxyribonucleic acid) that starts in one cell of the body at any time of life after an embryo is formed. All the cells that come from that cell will typically have the same mutation, which in some cases can lead to cancer. This kind of mutation is different from an inherited mutation, which is present at birth and found in all the cells of the body. Somatic mutations are not passed on to children.

Targeted Therapy

NCI (Cancer.gov)	ACS (Cancer.org)
A type of treatment that uses drugs or other substances to identify and attack specific types of cancer cells with less harm to normal cells. Some targeted therapies block the action of certain enzymes, proteins, or other molecules involved in the growth and spread of cancer cells. Other	Treatment with drugs that attack some part of cancer cells that's different from normal cells. Targeted therapies sometimes work when standard chemotherapy drugs don't, and they tend to have fewer side effects than chemotherapy drugs.

types of targeted therapies help the immune system kill cancer cells or deliver toxic substances directly to cancer cells and kill them. Targeted therapy may have fewer side effects than other types of cancer treatment. Most targeted therapies are either small molecule drugs or monoclonal antibodies.[30]	

Tumor Marker

NCI (Cancer.gov)	ACS (Cancer.org)
A substance found in tissue, blood, or other body fluids that may be a sign of cancer or certain benign (noncancerous) conditions. Most tumor markers are made by both normal cells and cancer cells, but they are made in larger amounts by cancer cells. A tumor marker may help to diagnose cancer, plan treatment, or find out how well treatment is working or if cancer has come back. Examples of tumor markers include CA-125 (in ovarian cancer), CA 15-3 (in breast cancer), CEA (in colon cancer), and PSA (in prostate cancer).[31]	Substances that can be found in the body (in the blood, urine, or other body fluids or tissues) when a person has cancer. These substances are usually made by cancer cells, but are sometimes by normal cells as well. Different types of cancer can have different tumor markers. For example, prostate-specific antigen (PSA) is a tumor marker for prostate cancer. Ideally, a tumor marker would always be found when a person has a certain type of cancer, and would never be found if a person didn't have cancer. But in reality tumor markers are rarely like that, because normal cells can often make them as well.

Tyrosine Kinase Inhibitor

NCI (Cancer.gov)	ACS (Cancer.org)
A substance that blocks the action of enzymes called tyrosine kinases. Tyrosine kinases are a part of many cell functions, including cell signaling, growth, and division. These enzymes may be too active or found at high levels in some types of cancer cells, and blocking them may help keep cancer cells from growing. Some tyrosine kinase inhibitors are used to treat cancer. They are a type of targeted therapy.[32]	Not found

[1] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=721252

[2] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=44643

[3] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=44918

[4] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=46086

[5] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=45618

[6] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=44397

[7] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=45693

[8] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=386201

[9] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=46128

[10] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=444994

[11] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=44593

[12] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=561401

[13] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=753865

[14] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=446543

[15] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=45983

[16] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=46365

[17] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=641114

[18] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=750798

[19] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=579630

[20] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=766166

[21] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=45065

[22] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=46063

[23] http://www.nature.com/jid/journal/v133/n8/full/jid2013248a.html

[24] http://www.ncbi.nlm.nih.gov/pubmed/21657839

[25] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=561717

[26] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=741769

[27] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=46092

[28] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=557359

[29] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=46586

[30] http://www.cancer.gov/publications/dictionaries/cancer-terms?cdrid=270742

[31] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=46636

[32] http://www.cancer.gov/publications/dictionaries/cancer-terms?CdrID=44833