



## Glossary

- **Albuterol:** (Beta 2 agonist or albuterol sulfate) The most commonly seen rescue or quick relief medication used to relieve asthma symptoms quickly by relaxing muscles around the airways. Also used before exercise to prevent asthma symptoms for people with exercise-induced asthma. Most often used as an inhaler or with a nebulizer.
- **Allele:** One of the variant forms of a gene. Different alleles may produce variation in inherited characteristics.
- **Allergen:** A substance which causes an allergic response in sensitive individuals. Allergens can be either natural (e.g., pollen, dust) or man-made (e.g., perfume, cleaning agents).
- **Alveoli:** Tiny air sacs where oxygen and carbon dioxide (a waste product) are exchanged. Oxygen goes to the body, and CO<sub>2</sub> is exhaled out.
- **Anti-inflammatory drugs:** Drugs that reduce the swelling and irritated tissues that are the symptoms and signs of inflammation.
- **Asthma:** A chronic, inflammatory disorder of the airways characterized by wheezing, breathing difficulties, coughing, chest tightness, and other possible symptoms. People with asthma have very sensitive airways that are constantly on the verge of over-reacting to asthma triggers.
- **Asthma Action Plan:** A written document that outlines an individual treatment plan for a person who has asthma; developed in talking with the health care provider, family member and caregivers. Effective action plans help patients control their asthma and live healthy active lives.
- **Asthma episode/attack/exacerbation:** A time when asthma symptoms flare up or intensify, requiring immediate adjustments in treatment and medication to get symptoms under control. Asthma episodes may occur suddenly, with few warning signs, or build slowly over a period of hours or even days.

- **Asthma management:** A comprehensive approach to achieving and maintaining control of asthma. It includes patient education to develop a partnership in management, assessing and monitoring severity, avoiding or controlling asthma triggers, establishing plans for medication and management of exacerbations, and regular follow-up care.
- **Atopy:** The increased likelihood (or possibility), usually because of genetics, that a person will develop allergic-type responses to common environmental allergens.
- **Attributable risk:** The excess risk of a specified health effect assumed to result from a specified exposure. In the case of complex disease, one can speak of the attributable risk associated with the genetic or environmental contributions.
- **Base pair:** Two bases, which form a “rung on the DNA ladder”. Bases are the “letters” (Adenine, Thymine, Cytosine, Guanine) that spell out the genetic code. Normally adenine pairs with thymine and cytosine pairs with guanine.
- **Bronchial tubes (bronchus):** The major airways of the respiratory system that carry air from the trachea (windpipe) to the microscopic air sacs (alveoli) in the lungs.
- **Bronchiole:** The smallest airways in the lungs.
- **Bronchoconstriction/ bronchospasm:** The tightening in the airways that occurs with asthma. Caused when the muscles around the bronchial tubes contract in response to specific triggers.
- **Bronchodilator drugs:** Drugs that relax muscles around the airways, which open the airways up. Some bronchodilators are used for quick relief of symptoms during an asthma attack. Others are taken every day to prevent symptoms.
- **Candidate gene:** A gene, located in a specific chromosomal region suspected of being associated with a disease, whose protein product is consistent with the known disease process.
- **Carrier testing:** Type of genetic testing that can tell individuals if they are carriers of a mutated gene. Carriers rarely develop the disease, but can pass on the mutated gene or the normal gene to their children (Example: cystic fibrosis).
- **Chromosome:** One of the threadlike "packages" of genes and other DNA in the nucleus of a cell. Humans have 23 pairs of chromosomes.

- **Complex trait:** A trait that results from the interaction of one or more genes and environmental factors (Example: asthma).
- **Confidentiality:** Acknowledgment that genetic information is sensitive and private and access should be limited to those authorized to receive it.
- **Controller medications:** The standard treatment of asthma for most patients who have “chronic” asthma and need daily medication. These medications provide “long-term relief” by making airways less sensitive which reduces symptoms before they even appear. Called a controller because it controls asthma and prevents asthma attacks.
- **Corticosteroid:** A type of medication used to reduce inflammation. These drugs mimic a substance produced naturally by the adrenal glands. In asthma, these drugs are often taken through an inhaler for long-term control. They may also be taken orally or given intravenously for a short time if asthma symptoms become out of control.
- **Dander:** Scaly or shredded dry skin that comes from animals or bird feathers. Dander may be a cause of an allergic response in some people.
- **DNA:** The organic molecules inside the nucleus of a cell that carries the genetic instructions for making living organisms.
- **Dry powder inhaler:** Similar to a metered dose inhaler except the medicine comes out of the DPI as a dry powder. The patient exhales out a full breath, places the lips around the mouthpiece, then quickly breathes in the powder.
- **Dyspnea:** Difficult breathing.
- **ELSI:** An abbreviation for the ethical, legal, and social implications of human genome research.
- **Eugenics:** An early twentieth century movement which aimed to improve the human race by preventing "unfit" people from reproducing (negative eugenics) and encouraging "fit" people to reproduce (positive eugenics). Eugenics was implemented in United States, the Nordic countries, and Germany. The eugenic philosophy is most widely known for the association with Nazi abuses and “better baby” and “fittest family” contests.

- **Exercise-induced asthma:** Asthma symptoms that appear during or following exercise. Symptoms may be minimal or severe enough to require emergency treatment. Some people who have chronic asthma have exercise as a trigger. Some people only develop bronchoconstriction (asthma symptoms) when they exercise.
- **Family health history:** Reflects a family's shared genetics, environment, culture, lifestyle, and behaviors. These combined factors affect a person's risk of developing disease.
- **First-degree relative:** An immediate family member such as a parent, sibling, or child.
- **Gene:** The basic unit of hereditary information passed from parent to offspring.
- **Gene-environment interaction:** The effects of one or more genes interacting with environmental factors in determining the occurrence of disease.
- **Gene-gene interaction:** The interaction of several different genes in the production of a disease.
- **Genetic counseling:** Provides patients and their families with education and information about genetic-related conditions and helps them make informed decisions.
- **Genetic discrimination:** Prejudice against those who have or are likely to develop an inherited disorder.
- **Genetic profile:** The particular arrangement of genes and markers in the DNA, unique to each individual.
- **Genetics:** The study of biological variation. Typically refers to a single gene and its effects.
- **Genetic screening:** Testing a *group* of people to identify individuals at high risk of having or passing on a specific genetic disorder.
- **Genetic testing:** Testing done by analyzing DNA to determine if an *individual* has certain mutations associated with an inherited condition.
- **Genome:** All of the DNA contained in an organism or a cell.
- **Genomics:** The study of all the genes and how they interact with each other and the environment.

- **Human Genome Project:** An international research project to map each human gene and to completely sequence human DNA, completed in 2003.
- **Informed consent:** Obtaining permission to do genetic testing. One must have knowledge of the risks, benefits, effectiveness, and alternatives to testing in order to understand the implications of genetic testing.
- **Inhaled corticosteroid:** Steroidal anti-inflammatory medication useful for people who have asthma. The medication is breathed in through the mouth into the lungs. Also called “ICS”. Not the same as anabolic steroids.
- **Inhaler/meter-dose inhaler (MDI):** A device used to deliver a variety of commonly prescribed asthma medications that help ease breathing by opening the airways.
- **Inherited:** Traits transmitted from parents to offspring.
- **Irritant:** Any substance that causes swelling of the respiratory system. An irritant may trigger asthma symptoms, but they may not be considered an allergen. Examples of irritants include tobacco smoke, chemicals, pesticides or air pollution.
- **Mucus:** Often called phlegm or sputum, this sticky fluid is produced by the membranes lining the airways. Exposure to certain triggers can increase mucus production. Excessive amounts of mucus make breathing more difficult.
- **Multifactorial:** A trait or disease resulting from interplay between multiple genes and environmental factors.
- **Mutation:** A permanent change in DNA. Some mutations can have no effect, can be beneficial, or cause harm to the organism.
- **Nebulizer:** A small, portable machine used to deliver certain asthma medications. It makes a mixture of liquid medicine and water into a mist that a person then inhales through a mask or a mouthpiece. They are often used for babies and children who are too small to coordinate using a MDI. They are also used for those having severe asthma symptoms, as it is easier to take in the medicine this way when having trouble breathing.

- **Newborn screening:** A public health screening program to screen every baby in the United States within the first few days of life, to look for inherited metabolic disorders.
- **Peak flow meter:** A small, portable hand-held device that measures how well the lungs are able to expel air, allowing people with asthma to detect airway narrowing and adjust medications accordingly.
- **Pedigree:** Diagram of a family's genealogy that shows family members' relationships to each other and how a particular trait or disease has been inherited.
- **Personalized medicine:** The development of drug therapies intended to treat people as individuals.
- **Pharmacogenomics:** The study of how an individual's genetic makeup affects their response to drugs.
- **Polymorphism:** A variation in the sequence of DNA among individuals found in at least 1% of the population.
- **Predictive testing:** Genetic testing to identify people who are at an increased risk for developing a certain type of disease or disorder.
- **Privacy:** The rights of individuals to keep personal information secure.
- **Pulmonary function testing (PFT's):** A series of tests used to determine whether a person has breathing problems, and precisely what those problems are. These test lung function and capacity. They do not hurt, as they involve tests that include holding your breath, blowing into a tube as hard as you can, and exercising while wearing a special mask.
- **Quick relief or rescue medications:** Medication taken to relieve asthma symptoms. Called "quick relief" because they can act immediately to reduce symptoms that appear suddenly.
- **Relative risk:** The chance of developing a specific disease as compared to the risk for another individual or group.
- **Second-degree relative:** A relative such as your grandparent, grandchild, uncle, aunt, niece, nephew, or half-sibling.
- **Single gene disorder:** A disorder caused by mutations within one particular gene (Example: Huntington's disease, Tay-Sachs).

- **Single Nucleotide Polymorphism (SNPs):** Pronounced “snips”. A DNA sequence variation that is one base long, and that occurs in at least one percent of the general population. SNPs account for much of the variety among humans.
- **Spacer:** A device that attaches to an inhaler (MDI) by a plastic chamber on one end and a mouthpiece on the other end. It is intended to help medicine from an MDI get into the lungs. A spacer works by holding the medicine in its chamber long enough for a person to inhale it in one or two slow deep breaths. Without a spacer much of the medicine in an inhaler “puff” is deposited on the tongue or in the back of the throat.
- **Spirometry:** Test for diagnosing asthma. A spirometer is an instrument that measures the maximum volume you can exhale after breathing in as much as you can. Small spirometers are available for home use, although peak flow meters are more appropriate for most people.
- **Third-degree relative:** A relative such as great grandparent, great uncle/aunt, or cousin.
- **Trigger:** Anything that causes asthma symptoms to worsen in a given person. Different things are triggers for different people. Some common triggers include exercise, cigarette smoke, pollen, dust, cold air, and upper respiratory infections.
- **Utah Genetic Testing Privacy Act:** This law protects Utah citizens from genetic discrimination in employment and some health insurance settings (To read the law visit <http://www.code-co.com/utah/code/03/26-45.htm>).
- **Wheezing:** A whistling sound that occurs when air moves through narrowed or tightened airways. May be heard on exhalation. Wheezing is a classic symptom of asthma.

**References:**

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