Glossary of terms

**A**
**Acceptable daily intake (ADI)**
The amount of chemical that, if ingested daily over a lifetime, appears to be without adverse effect.

**Acroparaesthesia**
Painful burning sensation in the hands and feet.

**Airway clearance techniques**Methods used to assist with secretion removal. There are several different techniques such as: chest physical therapy (CPT); positive expiratory pressure (PEP); high pressure PEP; flutter; autogenic drainage; and active cycle breathing.

**Albuterol**
An agent which acts relatively selectively on beta-2 adrenoceptors in the airways to cause bronchodilation. Its main clinical use is in asthma and is known in the UK as salbutamol, a brand name of this drug is Ventolin.

**Allele**
One of the variant forms of a gene at a particular locus, or location, on a chromosome. Different alleles produce variation in inherited characteristics such as hair color or blood type. In a dominantly inherited condition, if only one of the two genes is present, the disease/disorder occurs. In a recessively inherited condition, both genes must be present for disease to occur.

**Allergic bronchopulmonary aspergillosis (ABPA)**
An inflammatory and destructive disease of the bronchi and lungs due to the presence and growth of Aspergillus fumigatus.

**Alpha globin**
A blood protein found in the red blood cells. Alpha and beta globins combine to make haemoglobin, which carries oxygen.

**Alveoli**
The millions of tiny air sacs in the lungs, where oxygen and carbon dioxide are exchanged with the bloodstream.

**Amphotericin**
An antibiotic used to treat deep-seated fungal infections.

**Amino acids**
The building blocks of proteins, there are 20 naturally occurring amino acids.

**Aminoglycoside**
Broad spectrum antibiotics that are used to combat bacterial infections, can cause renal toxicity and hearing loss. Tobramycin and gentamicin are examples of these antibiotics and are often used by people with CF.

**Anaemia**
A condition where there are fewer red blood cells than average circulating in the blood stream. This may have a number of causes. See also 'Aplastic anaemia' and 'Sickle Cell Anaemia'.

**Angiokeratomas**
Red, raised lesions.

**Anthropometry**
Science and practice of measuring the human body and its parts.

**Antibiotic**
A drug which can destroy or inhibit growth of infectious organisms such as bacteria.

**Antibody**
A protein of the immune system, produced in response to an antigen (a foreign, often disease-causing, substance).

**Anticholinergic agents**
In airways these agents cause bronchodilation, an example of one of these agents is ipratroprium bromide.

**Antigen**
Any protein or polypeptide substance (such as a toxin or enzyme) capable of stimulating an immune response

**Antimicrobial**
Destroying or inhibiting the growth of microorganisms

**Antipseudomonal antibiotics**
Antibiotic drugs tending to destroy bacteria of the genus Pseudomonas.

**Aplastic anemia**
One type of anemia that occurs when the bone marrow produces too few of all three types of blood cells: red cells, white cells, and platelets. See also 'Anaemia'.

**Aplastic crisis**
This occurs when the body stops making new red blood cells and is usually caused by an infection. This is a life-threatening condition and must be treated immediately with blood transfusion and treatment of any accompanying infection.

**Apolipoprotein A1**
The protein component of serum lipoproteins (HDL cholesterol). Small proteins containing multiple copies of the kringle domain.

**Apolipoprotein B-100**
The protein component of LDL cholesterol.

**Arrhythmia**
Abnormal heart rhythm.

**Arthralgia**
Pain about/from a joint, not associated with swelling or redness.

**Arthritis**
The inflammation of joints.

**Ascites**
The accumulation of fluid in the peritoneal cavity.

**Aseptic necrosis**See 'Avascular necrosis'.

**Asymptomatic**
Absence of symptoms. A person is asymptomatic if he/she has no symptoms.

**Atelectasis**
A collapsed portion of the lung which does not contain air. This can be caused by excessive accumulations of mucous secretions, inhaled foreign bodies or bronchial cancers.

**Atheromatous disease**
A disease characterised by thickening and fatty degeneration of the inner coat of the arteries.

**Atopic**
Refers to a group of inherited diseases where there is often and inherited tendency to develop allergic conditions.

**Autogenic drainage**
A combination of breathing control and breaths at various lung volumes.

**Autosomal recessive**
A genetic trait or disorder which appears only when an individual inherits a pair of chromosomes, each containing the gene for the trait. One chromosome of the pair comes from the father and the other from the mother. Autosomal recessive disorders can occur only if both parents are carriers of the trait. Cystic fibrosis and sickle cell disease are inherited in this manner.

**Avascular necrosis**
When blood vessels supplying bone get blocked, resulting in tiny breaks within the bone which can eventually cause the bone to collapse and bone death to occur. Also called aseptic necrosis and osteonecrosis.

**B**
**Bacteria**
A single-celled organism. Bacteria are found throughout nature and can be beneficial or harmful.

**BAL**
See 'Bronchoalveolar lavage'.

**Beta-2 agonists**
Drugs which act on B2 receptors. These receptors are found in the airways and these drugs cause bronchodilation by relaxing the muscles in and around the airways that tighten during an asthma episode, e.g. albuterol.

**Beta globin**
A blood protein found in red blood cells that when mutated can lead to sickle cell disease. Beta and alpha globins combine to make haemoglobin, which carries oxygen.

**Biliary**
Of, relating to, or conveying bile.

**Bilirubin**
A pigment produced when the liver processes waste products. A high bilirubin level causes yellowing of the skin.

**Bisphosphonates**
A group of drugs that prevent the loss of calcium from bone and its transfer to the bloodstream. They are used to strengthen the bones in the treatment of Paget's disease (in which the bones become deformed and fracture easily), to lower high concentrations of calcium in the blood in patients with bone cancer, and to treat or prevent osteoporosis.

**Brasfield chest radiograph score**
A system for scoring chest X-rays in cystic fibrosis.

**Body mass index (BMI)**
A measure of nutritional status, BMI = weight in kg/(height in metres)2

**Bone marrow**
The soft, spongy tissue found in the centre of most large bones that produces the cellular components of blood: white cells, red cells and platelets.

**Broad spectrum antibiotics**
Antibiotics that are active against a wide variety of microorganisms.

**Bronchial drainage**
See 'Postural drainage'.

**Bronchial lavage**
A procedure in which small volumes of saline are rinsed through the lungs, followed by suction. It may be used in the treatment of CF to aspirate mucus secretions.

**Bronchiectasis**
Persistent and progressive dilation of bronchi (branches from the trachea which lead to the lungs) often as a consequence of inflammatory disease (lung infections).

**Bronchoalveolar lavage**
Saline is instilled by means of a bronchoscope which has been advanced as far as possible into the lungs and the fluid aspirated out from the alveoli.

**Bronchodilator**
A medication that acts to dilate (enlarge) the lumen of the airway to allow the unrestricted passage of air. Examples include: theophylline, aminophylline, adrenaline, Alupent, metaproterenol, isoproterenol, Ventolin, Proventil, bitolterol, salmeterol, pirbuterol and albuterol.

**Bronchospasm**
Spasmodic tightening or contracting of muscles surrounding and supporting bronchial tubes interfering with normal breathing and causing respiratory distress. Occurs in asthma attacks, in CF and other conditions.

**Button**
See 'Gastrostomy'.

**C**
**Cardiomyopathy**
A weakening of the heart muscle (myocardium), which usually causes inadequate heart pumping. It can be caused by viral infections, heart attacks, alcoholism, long-term and severe hypertension (high blood pressure).

**Cardiopulmonary failure**
Heart and lung failure.

**Carrier (as in genetics)**
An individual who possesses one copy of a mutant allele that causes disease only when two copies are present (an autosomal recessive disease). Although carriers not affected by the disease, two carriers can produce a child who has the disease.

**Catabolism**
The metabolic breakdown of complex molecules into simpler ones, often resulting in a release of energy.

**CBC**
See 'Complete Blood Count'.

**Cerebrospinal**
Of or relating to the brain and spinal cord or to these together with the cranial and spinal nerves that innervate voluntary muscles.

**Cerebrovascular**
Pertaining to the blood vessels of the cerebrum or brain.

**CF**
See 'Cystic Fibrosis'.

**CFTR**
See 'Cystic Fibrosis Transmembrane Conductance Regulator'.

**Chelation therapy**
The removal of heavy metal poisoning using agents which separate the metal from organs or tissues and bind it firmly with a new compound which can be eliminated from the body.

**Chest physiotherapy**
A treatment in diseases such as Cystic Fibrosis. Used to mobilise secretions and promote the removal of these secretions by an effective cough.

**Cholesterol**
A soft, waxy substance that is present in all parts of the body including the nervous system, skin, muscle, liver, intestines, and heart. It is made by the body and obtained from animal products in the diet.

**Cholestatic**
Tending to diminish or stop the flow of bile.

**Chromosome**
A structure found in the cell nucleus that contains the genes; chromosomes are composed of DNA and proteins. Each parent contributes one chromosome of each pair, so children get half of their chromosomes from their mothers and half from their fathers.

**Cirrhosis**
Widespread disruption of normal liver structure by fibrosis and the formation of regenerative nodules that is caused by any of various chronic progressive conditions affecting the liver.

**Ciprofloxacin**
A broad-spectrum quinolone antibiotic that can be given orally and is particularly useful against Gram-negative bacteria, such as Pseudomonas, that are resistant to all other oral antibiotics.

**CPT**
See 'Chest Physiotherapy'.

**Coagulopathy**
A disorder of the blood clotting (coagulation) system in which bleeding is prolonged and excessive.

**Collateral ventilation**
The entrance of air into alveoli through pulmonary alveolar pores and other pathways so that a lobule may remain aerated even though its bronchiole is obstructed. Also known as collateral respiration.

**Colistin**
An antibiotic effective against Pseudomonas administered in nebulised form or intravenously in the treatment of CF.

**Complete blood count**
A measurement of size, number, and maturity of the different blood cells in a specific volume of blood.

**Congenital**
Any trait or condition that exists from birth.

**Cor pulmonale**
Enlargement of the right ventricle of the heart due to resistance of the passage of blood through the lungs. Can often lead to right heart failure. This can be a major complication of chronic pulmonary diseases such as CF.

**Corpora cavernosa**
A mass of erectile tissue with large interspaces capable of being distended with blood; especially one of those that form the bulk of the body of the penis or of the clitoris.

**Corpus spongiosum**
The median longitudinal column of erectile tissue of the penis that contains the urethra and is ventral to the two corpora cavernosa.

**Crackles**
Also known as rales, a soft fine crackling sound heard in the lungs through the stethoscope. Made either by air passages and alveoli (air sacs) opening up during inspiration or by air bubbling through fluid. They are not usually heard in healthy lungs.

**Cryoprecipitate**
The precipitate that forms when plasma is frozen and then thawed, particularly rich in fibronectin and blood clotting Factor VIII.

**Cystic fibrosis**
A genetic disease with symptoms that usually appear shortly after birth. They include breathing difficulties and respiratory infections due to accumulation of sticky mucous problems with digestion and excessive loss of salt in sweat. The disease affects many organs including the lungs, gastrointestinal tract, pancreas and liver.

**Cystic fibrosis transmembrane conductance regulator (CFTR)**
A protein, involved in the movement of salt across cell membranes, which is lacking or does not function normally in people with cystic fibrosis.

**Cytokine**
Protein molecules, released by cells when activated by antigen, that are involved in cell-to-cell communications, acting as enhancing mediators for immune responses through interaction with specific cell-surface receptors on leucocytes.

**Cytopenia**
A reduction in the number of cells circulating in the blood.

**Cytostatic**
Tending to retard cellular activity and multiplication.

**D**
**Dactylitis**
Painful swelling of the feet and hands during the first several years of life of children with sickle cell anaemia.

**Deferiprone**
A drug which acts by combining with the excess iron in the body to form a complex which is excreted in the urine. This over a period of time leads to reduction in the iron overload in the body. Also known as L1.

**Desferrioxamine**
A drug that combines with iron in body tissues and fluids and is used to treat iron overload (including that resulting from prolonged or constant blood transfusion, as for thalassaemia), diseases involving iron storage in parts of the body, and for the diagnosis of such diseases.

**Diabetes (Diabetes mellitus)**
A pancreatic disorder that causes abnormal insulin production. This affects the body's ability to utilise sugar and other food substances and is usually treated by diet modification (restricted sugar intake) and use of insulin. In cystic fibrosis, disruption of pancreatic function sometimes leads to a form of the disease called cystic fibrosis related diabetes mellitus (CFRDM).

**Distal intestinal obstruction syndrome**
Occurs in CF and a form of partial intestinal obstruction which presents with a history of cramping pain and changes in stool pattern in absence of other acute GI symptoms. Usually responds to medical treatment.

**DNA**
Deoxyribonucleic acid, the chemical coding for a gene. DNA determines the "genetic message" within each cell, organ, and organism.

**Dyspnoea**
Shortness of breath, difficult or laboured breathing.

**E
Elective**
Planned as opposed to emergency treatment.

**Embolisation**
The blocking or obstruction of a blood vessel or organ by the lodgement of a material mass (as an embolus).

**Endemic**
Widely prevalent in a particular region.

**Endobronchial**
Within the bronchi or bronchial walls.

**Endocytosis**
The process by which materials enter a cell without passing through the cell membrane, the plasma membrane folds inward to bring substances into the cell.

**Enteral nutrition**
Nutrition provided through a tube or catheter that delivers the nutrients directly into the stomach.

**Enteric-coated**
A special coating applied to tablets or capsules which prevents release and absorption of their contents until they reach the intestines.

**Enzymes**
Substances which help produce and/or accelerate certain chemical processes in the body, such as breaking down of foods during digestion. Individuals with cystic fibrosis may need pancreatic enzyme replacements to help with proper digestion of food.

**Epigastric**
Of, or relating to, the anterior walls of the abdomen.

**Epithelial tissue**
Cells which form a lining on the outside of the body and on the inside of the respiratory and digestive tracts and other organs.

**Ergometry**
Any method of measuring the amount of work done by an organism, usually during exertion.

**ERV**
See 'Expiratory Reserve Volume'.

**Erythema**
Abnormal redness of the skin or other surface due to capillary congestion (as in inflammation).

**Erythrocytapheresis**
Automated red cell transfusion.

**Erythrocytes**
Red blood cells containing haemoglobin, the substance which carries oxygen in the bloodstream.

**Essential thrombocythaemia**
A condition characterised by the production of large numbers of abnormal platelets. Symptoms include haemorrhage, blood clots and enlargement of the spleen.

**Exacerbation**
Aggravation of symptoms or increase in the severity of a disease.

**Expectorate**
To spit; to eject saliva, mucus, or other fluid from the mouth.

**Expiratory reserve volume (ERV)**
This represents the reserve volume of air you can exhale after you have exhaled normally during a resting respiration. Again, your body can call upon this reserve to increase the amount of air exchange.

**Exocrine glands**
Glands which secrete substances through ducts to surrounding surfaces. Includes sweat, salivary and tear glands, as well as the mucous glands in the digestive, respiratory, and genitourinary systems. These glands are greatly affected in CF. Their ducts may be obstructed by mucus.

**F
Factor VIII**
A soluble blood protein that forms part of the cascade leading to blood clotting. The gene coding for factor VIII is found on the X chromosome in humans. A mutation in this gene in males leads to the disease Haemophilia A, in which a person's blood does not clot normally.

**Factor IX**
A soluble blood protein that forms part of the cascade leading to blood clotting. The gene coding for factor VIII is found on the X chromosome in humans. A mutation in this gene in males leads to the disease Haemophilia B, in which a person's blood does not clot normally.

**False positive result**
A person is identified by a test as having a condition/abnormality when they do not.

**False negative result**
A person is identified by a test as not having a condition/abnormality, when in fact they do.

**Familial hypercholesterolaemia (FH)**
An inherited disorder caused by any one of over 300 mutations (defects) in the gene for the low density lipoprotein (LDL) receptor. It is characterised by a total blood cholesterol level of above 7.5 mmol/l or a blood concentration of LDL cholesterol of above 4.9mmol/l (hypercholesterolaemia) combined with the presence of xanthomatosis and premature ischaemic heart disease in the patient or a close relative.

**FEF25-75%**
See 'Forced Expiratory Flow Rate'.

**Fetal Hemoglobin (Hgb.F)**
A kind of hemoglobin usually present during fetal (intrauterine) life, which has a different chemical structure from normal adult hemoglobin. After birth, the fetal hemoglobin in the red blood cells is gradually replaced by the adult type of hemoglobin, this process is usually complete during the first 6 months of life.

**FEV1**
See 'Forced Expiratory Volume in one second'.

**Forced expiratory flow rate**
The average flow rate measured over the middle half of the expiration.

**Forced expiratory volume in one second**
Often abbreviated to FEV1, it is the volume of air one breathes out forcefully in one second after taking a deep breath in. A standard measure of lung function and an efficacy parameter used in CF studies. It is a good predictor of mortality in CF patients.

**Forced vital capacity**
The total amount of air exhaled forcefully after a deep inspiration.

**FRC**
See 'Functional Residual Capacity'.

Functional Residual (Reserve) Capacity
The amount of air remaining in the lungs after a normal exhalation. It cannot be measured directly. Functional Reserve Capacity is the sum of the Expiratory Reserve Volume and Reserve Volume.

**FVC**
See 'Forced Vital Capacity'.

**G
Galactosemia**
A genetic disorder resulting in the impairment of transformation of the simple sugar galactose to glucose.

**Gastrointestinal**
Concerning the digestive system, e.g. the stomach, bowel.

**Gastrostomy**
A device permanently placed in the stomach to facilitate supplemental feedings; MIC and Bard buttons are two kinds of buttons.

**Gaucher disease**
An inherited metabolic disorder in which harmful quantities of a fatty substance called glucocerebroside accumulate in the spleen, liver, lungs, bone marrow, and, in rare cases, the brain.

**Gene**
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.

**Gene therapy**
A form of therapy where a normal, functioning gene is introduced into a cell in which that gene is missing or defective.

**Gene transfer**
Insertion of unrelated DNA into the cells of an organism. There are many different reasons for gene transfer: for example, attempting to treat disease by supplying patients with therapeutic genes.

**Genetic disorder**
A condition, which is the result of alterations in the genetic make-up of an individual. They may be the direct consequences of defects in single genes (mutations); or in whole chromosomes, parts of which may be lost, duplicated or misplaced; or from the interaction of multiple genes and external factors.

**Genome**
All the DNA contained in an organism or a cell, which includes both the chromosomes within the nucleus and the DNA in mitochondria.

**Genotype**
The genetic identity of an individual that does not show as outward characteristics.

**GI**
See 'Gastrointestinal'.

**Glucocorticoids**
A group of anti-inflammatory drugs that are related to cortisol, a natural steroid hormone produced by the body.

**H**
**H2 receptor antagonists**Drugs which act by blocking the H2 receptor. In the GI tract the effect of these drugs is to lower gastric acid secretion.

**Haematocrit**
The hematocrit is the percent of whole blood that is comprised of red blood cells. The hematocrit is a compound measure of red blood cell number and size.The hematocrit is almost always included as part of the full blood count (FBC).

**Haemoglobin**
A protein found in the red blood cell, two beta, two alpha and four hemes combine to make one haemoglobin molecule. Haemoglobin carries and delivers oxygen to the cells.

**Haemoglobin type and description**
Hb A: Usual haemoglobin, also called adult haemoglobin.
Hb AA: The inheritance of Hb A from both parents.
Hb S: Sickle Haemoglobin, a beta chain variant.
Hb AS: Sickle cell trait, also known as carrier state. The inheritance of Hb A from one parent and Hb S from the other parent.
Hb SS: Sickle cell anaemia. The inheritance of Hb S from both parents, also known ashomozygous for HbS.
Hb CHaemoglobin C, another beta chain variant.
Hb AC: Hb C trait. The inheritance of Hb A from one parent and Hb C from the other parent.
Hb SC: SC disease. The inheritance of Hb S from one parent and HbC from the other parent, a compound heterozygote condition.
Hb CC: Hb C disease. The inheritance of Hb C from both parents.
Hb A Beta-Thal:Beta-Thalassaemia trait. The inheritance of Hb A from one parent and Hb Beta-Thal trait from the other parent.
Hb S Beta-Thal: Sickle Beta-Thalassaemia. The inheritance of Hb S from one parent and Beta-Thal trait from the other parent, a compound heterozygote state.
Hb A2: An adult haemoglobin usually only present in small amounts but present in increased amounts in Beta-Thalassaemia trait. Quantitation of this haemoglobin can therefore be useful in the diagnosis of this condition.
Hb FFetal haemoglobin which can be raised in Beta-Thal trait and sickle cell disease.

**Hemoglobin C trait (AC)**
The inheritance of one gene for the usual hemoglobin (A), and one gene for hemoglobin (C). A person who has hemoglobin C Trait (AC) is a carrier of the hemoglobin C gene, and is not affected by the gene.

**Haemoglobinopathy**
A group of inherited disorders involving abnormal production or structure of the haemoglobin chains. These disorders include the family of sickle cell disease, haemoglobin C disease, haemoglobin S-C disease, sickle cell anemia (SS), as well as as well as the thalassaemias.

**Haemolysis**
Breaking of the red cell membrane causing release of haemoglobin.

**Haemophilia**
An inherited bleeding disorder (also coagulation disorder) caused by low levels, or absence of, a blood protein that is essential for clotting; haemophilia A is caused by a lack of the blood clotting protein factor VIII; haemophilia B is caused by a deficiency of factor IX.

**Haemoptysis**
The expectoration or spitting up of blood or bloody mucus from the lungs, throat, or mouth.

**Haemostasis**
The arrest of bleeding, either by vasoconstriction, coagulation or by surgical means.

**Hydroxyurea**
Hydroxyurea is in a class of drugs known as urea derivatives. In sickle cell anemia, hydroxyurea decreases the episodes of painful crisis by decreasing the sickling of red blood cells.

**Hyperglycemia**
High blood sugar. Can occur in people with CF as a result of diabetes mellitus. This condition is easily controlled through use of insulin.

**Hypertonic saline**
Water with a concentration of more than 0.9% (which is isotonic) salt.

**Hypohidrosis**
A substantially decreased ability to sweat.

**Hypopnea**
Breathing that is shallower, and/or slower, than normal.

**Hypotension**
An abnormally low blood pressure.

**Hypotonia**
Low muscle tone.

**Hypoxia**
Lack of oxygen in the blood which can cause discolouration of the skin or mucuos membranes. May occur in lung diseases such as CF.

**Hypoxemia**
A lack of oxygen in the blood - Coalition for pulmonary fibrosis.

**I
IC**
See Inspiratory Capacity

**Immunosuppression**
The process of inhibiting a normal immune response with the use of drugs, biological agents, or chemical agents; commonly used in association with tissue transplantation or to control autoimmune diseases.

**In vitro**
Outside the living body and in an artificial environment

**Inborn errors of metabolism**Inherited diseases resulting from alterations in genes that code for enzymes.

**Infarct**
An area of tissue death due to a local lack of oxygen.

**Inhaled corticosteroids**
Inhaled corticosteroids are steroid medicines given into the lungs through an inhaler device, frequently used to treat asthma.
Nasal corticosteroids are steroid medications sprayed into the nose to help relieve the symptoms of conditions such as hay fever.

**Inspiratory capacity**
Equals the total amount of air you can inhale from a resting exhalation. Another way of looking at inspiratory capacity is:
Tidal Volume + Inspiratory Reserve Volume = Inspiratory Capacity

**Inspiratory residual (reserve) volume**
Represents the amount of air you can breathe in after a resting inspiration. This is the amount of air volume your body can call upon when a deeper breath is needed during exercise or when your body is under stress. Your body increases respiratory volume when it needs more oxygen and needs to blow off excess carbon dioxide.

**Intravenous**
Situated within, performed within, occurring within, or administered by entering a vein

**Iron chelating agents**
Chemical compounds which form complexes by binding metal ions. Some chelating agents, including desferrioxamine and penicillamine, are drugs used to treat metal poisoning: the metal is bound to the drug and excreted safely. Chelating agents often form the active centres of enzymes.

**IRV**
See 'Inspiratory Residual (Reserve) Volume'.

**Ischaemia**
A low oxygen state, usually due to inadequate blood flow, may be the result of obstruction or low perfusion leading to low oxygen levels in the tissue.

**Ischaemic heart disease**
Where the heart is damaged by inadequate blood flow.

**Isotonic saline solution**
A solution of sodium chloride and distilled water having the same concentration of solutes as the blood.

**J**

**K**

**L
Laryngitis**
Laryngitis is an inflammation of the larynx (voice box), may be caused by infection.

**Lesion**
An abnormal structural change in body tissues or organs. For example, the scar tissue in the lungs of a child with CF.

**Leucocyte, Leukocyte**
White blood cell, involved in body defence systems.

**Lipids**
Fatty molecules which are used by the body for energy. They are an important part of cell structure. People with CF with pancreatic insufficiency lack digestive enzymes responsible for the breakdown of fats into lipids.

**Liposomes**
A synthetic membrane vesicle made from phospholipids and used for in vitro study of membrane-defined events such as transport, or for the delivery of substances to a cell.

**Longitudinal study**
Measuring the performance of a sample on more than one occasion over a period of time.

**Lower respiratory tract**
The part of the respiratory tract situated below the vocal cords.

**LRT**
See 'Lower respiratory tract'.

**Lyophilize**
To isolate a solid substance from solution by freezing the solution and evaporating the ice under vacuum.

 **M
Macrolides**
A group of antibiotics including azithromycin, clarithromycin and erythromycin. Erythromycin has a similar spectrum to penicillin and may be used as an alternative to this drug. Azithromycin and clarithromycin have a broader spectrum.

**Maxillary sinus**
An air filled cavity within the maxilla (bone that forms the face and upper jaw). The maxillary sinus is located just below the bony prominence of the cheek.

**Meconium**
The first stool of a newborn infant, usually passed within a few hours after birth. Contains mucus and other secretions of intestinal glands.

**Meconium ileus**
Obstruction of the intestines of a newborn infant with abnormally thick meconium. The earliest symptom of CF, it presents as intestinal obstruction in the neonatal period. It occurs in seven to 10 percent of people with CF.

**Methicillin-resistant *Staphylococcus aureus* (MRSA)**
A bacterial strain that is resistant to methicillin and most other antibiotics.

**MIC (minimum inhibitory concentration)**
The lowest concentration of an antibiotic required to inhibit the growth of an organism in vitro.

**Morbidity**
Illness.

**Mortality**
Death.

**Mucoid**
Pertaining or relating to, or resembling mucus.

**Mucociliary clearance**
Mucociliary clearance of the respiratory tract is an important defence mechanism against inhaled pathogens. Cilia, which line both the upper and lower airways, are covered by a thin layer of mucus, and beat rapidly in a co-ordinated fashion propelling particles trapped in the mucus layer to the pharynx. Defective mucociliary clearance predisposes the respiratory tract to recurrent infection.

**Mucolytic agents**
Drugs which are used to break down mucus so that it can be coughed up.

**Mucosal oedema**
Accumulation of fluid and swelling of any membrane or lining which contains mucous secreting glands.

**Mucoviscidosis**
Term once used for cystic fibrosis. Still used in Europe. Mucoviscidosis was coined in 1944 to describe the abnormally viscid (thick and sticky) mucous secretions characteristic of CF.

**N
Nasal polyps**
Small growths of swollen mucous membrane which project into the nasal passages. Common in children with CF, they maybe multiple or recurrent.

**Nasogastric feeding**
Feeding through a tube inserted via the nose which reaches down into the stomach.

**Nebuliser**
A device used for generating a mist or aerosol from a solution. In the treatment of CF, nebulisers may be used for delivering aerosol antibiotics, mucolytic or other drugs.

**Neonate**
A newborn less than or equal to 28 days of age.

**Nephrotoxicity**
Something which damages the kidneys.

**Neuropathy**
Any disease of the peripheral nerves, usually causing weakness and numbness.

**Neutropenia**
Abnormally low number of leukocytes (mainly neutrophils) in the blood, most commonly due to a decreased production of new cells in conjunction with various infectious diseases, as a reaction to various drugs or other chemicals, or in response to irradiation.

**Neutrophil**
A variety of granulocyte (a type of white blood cell) distinguished by a lobed nucleus. It is capable of ingesting and killing bacteria and provides an important defence against infection.

**Non-invasive**
Procedure which does not require incision/invasion into the body or the removal of tissue.

**O
Ocular**
Having to do with the eye

**Omega-3 fatty acids - DHA/EPA -**
A type of fatty acid found in fish and marine oils which provides the health benefits of reduced risk of cardiovascular disease and improved mental and visual function.

**Opiate**
A drug or therapy containing or derived from opium.

**Osteomyelitis**
Infection of bone, it may remain localised or may spread through the bone.

**Osteopenia**
Reduction in bone volume to below normal levels especially due to inadequate replacement after the normal breakdown of bone.

**Osteoporosis**
A condition that affects especially older women and is characterized by decrease in bone mass with decreased density and enlargement of bone spaces producing porosity and brittleness.

**Ototoxicity**
Impacting on either hearing or balance or both.

**Oximetry**
Oximetry measures the percentage of hemoglobin saturated with oxygen by passing specific wavelengths of light through the blood. It includes non-invasive oxygen monitoring by pulse oximetry.

**P
Pancreas**
A tongue-shaped glandular organ lying below and behind the stomach. It secretes the hormones insulin and glucagon (both regulate blood sugar), in addition to pancreatic enzymes involved in the digestion of fats and proteins in the small intestine.

**Parenteral nutrition**
Nutrition taken intravenously (bypassing the digestive tract) in a hospital, nursing home or patient's home. You may also see the terms TPN (total parenteral nutrition) or HA (hyperalimentation) used.

**Paresthesias**
Abnormal touch sensations, such as burning or prickling, that occur without an outside stimulus.

**Parietal cell**
One of the cell's of the gastric glands, it secretes hydrochloric acid that reaches the lumen of the gland through fine intracellular and intercellular canals (canaliculi).

**Pathogen**
A specific causative agent (as a bacterium or virus) of disease.

**Peak flow meter**
A portable, inexpensive, hand-held device used to measure the flow of air during maximal expiration.

**Peak expiratory flow rate**
The greatest rate of airflow that can be achieved during forced expiration beginning with the lungs fully inflated.

**PEP device**
See 'Positive expiratory pressure device'.

**PFT**
Pulmonary Function Test.

**Pharyngitis**
Pharyngitis is defined as inflammation of the mucous membranes and submucosal structures of the pharynx. There are many agents which cause pharyngitis, the most common being viruses. Common signs and symptoms include sore throat, fever, headache, and nausea and vomiting.

**Phenotype**
The characteristics displayed by an individual under a particular set of environmental factors, regardless of the actual genotype of the individual.

**Phenylketonuria**
An inherited autosomal recessive disorder that results from a lack of the enzyme phenylalanine hydroxylase. This leads to accumulation of the amino acid phenylalanine that can impair development of the nervous system and cause neurodevelopmental impairment.

**Phenylalanine**
One of the amino acids which the body cannot manufacture itself, but must acquire from food., abundant in meats and cheese. Phenylalanine is a precursor of tyrosine and together they lead to the formation of adrenaline which is converted into a brain chemical which transmits nerve impulses and is used to manufacture noradrenaline which in turn promotes mental alertness, memory, elevates mood and suppresses the appetite.

**Phrynoderma**
A form of follicular hyperkeratosis associated with some micronutrient, e.g. vitamin A deficiencies.

**Phytomedicines**
Therapeutics based only on plant material, either from the complete plant or extracts, used for treatment purposes.

**PKU**
See 'Phenylketonuria'.

**Platelet**
Blood cell with no nucleus that helps to control bleeding by causing clotting; also called thrombocyte.

**Pneumothorax**
A condition in which air has entered and expanded the normally closed pleural space, driving pleural pressure up toward atmospheric pressure, and resulting in partial or complete collapse of the lung. Can be a complication of CF.

**Pneumococcal prophylaxis**
Protective or preventive treatment against a bacterium of the genus Streptococcus (S. pneumoniae) that causes an acute pneumonia involving one or more lobes of the lung.

**Positive expiratory pressure (PEP) device**Devices which provide a constant back pressure to the airways during expiration.

**Portal hypertension**
Any increase in the portal vein (in the liver) pressure due to anatomic or functional obstruction (for example alcoholic cirrhosis) to blood flow in the portal venous system.

**Postural drainage**The use of gravity assisted positioning to enable clearance of secretions. Also called bronchial drainage.

**Prednisolone**
A glucocorticoid with the general properties of the corticosteroids.

**Priapism**
This occurs in Sickle Cell Disease. The continued erection (generally painful) of the penis without sexual desire, occurs when sickle cells block blood circulation in the penis. Stuttering priapism is defined as recurrent episodes of prolonged erections.

**Procoagulant factor VIII**
A coagulation (clotting) factor. Classic haemophilia (haemophilia A) is due to a congenital deficiency in the amount (or activity) of factor VIII. Factor VIII is also known as antihemophiliac factor (AHF) or antihemophiliac globulin (AHG). See also Factor VIII.

**Prophylactic**
Referring to prevention, such as drugs used to prevent disease or complications.

**Prostaglandin E2 analogue**
A hormone-like substance which reduces the production of stomach acid and protects the stomach lining, e.g. misoprostol.

**Proteinuria**
The presence of protein in the urine.

**Protease**
An enzyme that digests proteins.

**Proton pump inhibitors**
A group of anti-ulcer medications which work by binding to an enzyme which is found on the secretory surface of parietal cells. It thereby inhibits the final transport of hydrogen ions (via exchange with potassium) into the gastric lumen and reduces acid secretion, e.g. omeprazole.

***Pseudomonas aeruginosa***
Bacteria (with a single circular chromosome) which frequently colonise the lungs of people with CF and are a major cause of respiratory infections. Prefer moist environments. Produce a bluish and greenish pigment and have a distinct "fruity" odour. The infection is harmful because the bacteria create toxins and enzymes which destroy the host tissue and cause the complications which are usually associated with cystic fibrosis.

**Pulmonary**
Relating to the lungs.

**Pulmonary function tests**
Test procedures used to evaluate lung function. Along with patient history and physical examination, pulmonary function tests are used to make the diagnosis, plan therapy, and determine prognosis. Can be used with adults and with children at least five to six years of age. Pulmonary function tests are used to measure: flows (of air) and timed volumes; tidal volume (amount of air entering and leaving the lungs during natural respiration); maximum voluntary ventilation (amount of air forcefully expired during one minute); residual volume of air remaining in the lungs after maximum voluntary expiration); total lung capacity (total amount of air in the lungs after maximum inspiration); and vital capacity (maximum amount of air expired after a full inspiration.) All pulmonary function tests are defined separately in this glossary.

**Q**

**R
Rales**
See 'Crackles '.

**Recombinant DNA molecule**
Recombinant DNA is DNA that has been created artificially. DNA from two or more sources is incorporated into a single recombinant molecule.

**Red blood cell dehydration**
Loss of water from red blood cells.

**Respiratory rate**
Number of breaths per minute.

**RNA**
Ribonucleic acid. Molecules of nucleic acid which are formed in the cell's nucleus (as directed by DNA). RNA is responsible for assembling proteins.

**Residual volume**
The amount of air left in the lung after you blow out as much air as possible.

**RV**
See 'Residual Volume'.

**S
Saline**
A mixture of salt and water.

**SCD**
See 'Sickle Cell Disease'.

**Screening**
Examination of a group of usually asymptomatic individuals to detect those with a high probability of having a given disease, typically by means of an inexpensive diagnostic test.

**Sepsis**
The presence of organisms in the blood.

**Serum ferritin**
An indirect measure of iron in the blood.

**Sequelae**
A condition which follows as a consequence of a disease or another condition.

**Serum**
The clear, liquid portion of blood which separates in the clotting of blood.

**Shwachman-Kulczycki score**A general score of clinical severity in cystic fibrosis assessed at annual review. Score separately each of the 4 areas (general activity, physical examination, nutrition and X-ray findings).

**Skin excoriation**
Abrasion or wearing off the skin.

**Sickle cell anaemia**
A condition caused by a mutation in the beta globin gene leading to production of abnormal haemoglobin. This causes the red blood cells to take on a distorted 'sickle' shape which severely reduces oxygen carrying capacity. Sickle cell anaemia is inherited from both parents as an autosomal recessive trait.

**Sickle cell disease (SCD)**
An inherited disorder of the red blood cells in which one gene is for sickle haemoglobin (S), and the other gene is for another unusual haemoglobin such as S, C, Beta Thalassaemia. etc. The following are examples of sickle cell disease: SS, SC, SD, SßThal, etc. It is seen most commonly in people of African, Mediterranean, Arabian and Indian ancestry. The family of clinically significant disorders including SS and compound heterozygote conditions.

**Sickle cell retinopathy**
People with sickle cell disease may develop sickle cell retinopathy, which can occasionally lead to blindness. This condition manifests slowly and rarely occurs before the age of ten years. Sickle cell retinopathy is more common in those with Hemoglobin SC disease than SS.

**Sickle cell trait (AS)**
The inheritance of one gene for the usual haemoglobin (A) and one gene for sickle haemoglobin (S). A person who has sickle cell trait (AS) is a carrier of the sickle gene, does not have the disease, does not have painful episodes, and is rarely affected by the sickle haemoglobin.

**Splenectomy**
The surgical removal of the spleen.

**Splenic sequestration**
Young children with SCD may have episodes of blood pooling in the spleen dropping Hb ³2, which may be life threatening.

**Spirometer**
Instrument used to measure lung air volumes and flow rates.

**Spirometric lung function**The vital capacity of the lungs, or the volume of air which can be expelled from the chest after the deepest possible inspiration.

**Sputum**
Mucus and other materials coughed up from lungs. Phlegm.

***Staphylococcus aureus***
Bacterial infection which most commonly inhabits the human nasal cavity. They occur in regular, grapelike clusters. Characteristic symptoms are pus filled inflammatory regions called abscesses. This same bacterium also causes Toxic Shock Syndrome and some types of food poisoning. In CF, "staph" commonly causes respiratory infections. Can often be effectively treated with antibiotics.

**Steatorrhea**
Faecal matter that is frothy, foul-smelling and floats because of a high fat content. In cystic fibrosis, this is caused by blockage of pancreatic ducts. Fats are not properly broken down and absorbed and are therefore excreted in the stools.

**Stroke**
A sudden attack of weakness or paralysis affecting a part (generally one side) of the body that lasts ³ 24 hours. It is usually caused by a disruption of blood flow to a region of the brain. A stroke can range in severity from those that recover to severe paralysis, coma or death.

**Sweat electrolytes**
Chemical ions contained in sweat. Some electrolytes (sodium and chloride) are elevated in most people with CF.

**Sweat test**
The diagnostic test for CF. Measures the concentration of salt (sodium and chloride) in sweat which are elevated in CF.

**T
Tachycardia**
The excessive rapidity in the action of the heart, the term is usually applied to a heart rate above 100 per minute.

**Tachypnea**
An abnormally rapid respiratory rate.

**Taurine**
Is one of the most abundant amino acids in the body. It is found in the central nervous system, skeletal muscle and is very concentrated in the brain and heart. Taurine seems to inhibit and modulate neurotransmitters in the brain.

**Thalassaemias**
A group of inherited blood disorders resulting from reduced or absent production of globin chains for the haemoglobin molecule in red blood cells. This leads to mild to severe anaemia. Other symptoms may include an enlarged spleen and anormalities in the bone marrow. Severe cases of the disease occur in patients who have both Beta globin genes mutated. Carriers (trait or Heterozygotes) are only mildly affected or have no symptoms at all. The Thalassaemias are widespread in Mediterranean countries, Asia and Africa.

**Thalassaemia intermedia**
When two Beta Thalassaemia genes have been inherited but the patient only requires infrequent transfusions.

**(Beta) Thalassaemia major**
An inherited disease with little or no beta haemoglobin production that requires lifelong red cell transfusions.

**Thalassaemia minor (trait)**
Reduced production of haemoglobin chains that is sometimes inherited along with sickle haemoglobin to produce sickle-beta thalassaemia. Having the trait by itself, is not harmful.

**Tidal volume**
While at rest our normal breath is called the Tidal Volume. Stress, exercise, and illness (including asthma) will cause this volume to increase or decrease.

**TLC**
See 'Total Lung Capacity'.

**Total lung capacity**
The total maximum amount of air your lungs can hold. It is the total of all lung volumes or lung capacities.

**Transcranial doppler ultrasonography**
A technique used to measure the velocity of blood flow in the arteries supplying the brain

**Transcutaneous**
Entering through the dermis or skin, as in administration of a drug applied to the skin in ointment or patch form.

**Triglyceride**
A combination of glycerol and three fatty acids (i.e., body fat).

**TV**
See 'Tidal Volume'.

**Tyrosine**
One of the twenty amino acids directly coded in proteins, can normally be synthesised from phenylalanine.

**U
Ursodeoxycholic acid**
Used in the treatment of gallstones. It is taken by mouth to dissolve the gallstones It is also used to help prevent gallstones in patients who are on rapid weight-loss programs.

**V
Vasodilatation**
Widening of the blood vessels.

**Vaso-occlusion**
Blockage of arteries.

**Venous access devices**
Catheters placed directly into the venous system for infusion therapy and/or phlebotomy.

**VC**
See 'Vital Capacity'.

**Venipuncture**
Collection of blood specimen from a vein for laboratory testing.

**Viral vectors**
Gene delivery systems that use viruses to transfer gene to cells.

**Vital capacity**
Represents the total amount of air exhaled from a maximal inhalation to a maximal exhalation, i.e.TLC & RV

**W**

**X
Xanthomatosis**
Deposits of cholesterol in the tendons and skin.

**Xerosis**
Abnormal dryness, as of the skin or eyeball.

**Y**

**Z**