Geneticheck - Genetic Report

Gene

CFTR

Associated Diseases

Idiopathic Bronchiectasis
Aquagenic Palmoplantar Keratoderma
Cystic Fibrosis
Vas Deferens, Congenital Bilateral Aplasia Of
Pancreatitis, Hereditary
Cystic Fibrosis
Bronchiectasis With Or Without Elevated Sweat Chloride 1
Congenital Bilateral Absence Of Vas Deferens
Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation
Hereditary Chronic Pancreatitis

Phenotype

Chronic lung disease
According to the definitions of the American and British Thoracic Societies, including pulmonary functional tests, X-rays, and CT scans for items such as fibrosis, bronchiectasis, bullae, emphysema, nodular or lymphomatous abnormalities.

Obstructive azoospermia
Absence of any measurable level of sperm in his semen, resulting from post-testicular obstruction or retrograde ejaculation. This can be differentiated from obstructive azoospermia on the basis of testicular biopsy.

Jaundice
Yellow pigmentation of the skin due to bilirubin, which in turn is the result of increased bilirubin concentration in the bloodstream.

Chronic bronchitis
Chronic inflammation of the bronchi.

Pulmonary fibrosis
Replacement of normal lung tissues by fibroblasts and collagen.

Elevated circulating C-reactive protein concentration
An abnormal elevation of the C-reactive protein level in the blood circulation.

Absent vas deferens
Aplasia (congenital absence) of the vas deferens.

Acute infectious pneumonia
Acute inflammation of the lung due to an infection.

Orthokeratotic hyperkeratosis

A form of hyperkeratosis characterized by thickening of the cornified layer without retained nuclei.

Recurrent respiratory infections
An increased susceptibility to respiratory infections as manifested by a history of recurrent respiratory infections.

Reduced FEV1/FVC ratio
Abnormally low FEV1/FVC (FEV1 - forced expiratory volume in 1 second; FVC forced vital capacity).

Nasal polyposis
Polypoidal masses arising mainly from the mucous membranes of the nose and paranasal sinuses. They are freely movable and nontender overgrowths of the mucosa that frequently accompany allergic rhinitis.

Pancreatic pseudocyst
Cyst-like space not lined by epithelium and contained within the pancreas. Pancreatic pseudocysts are often associated with pancreatitis.

Systemic lupus erythematosus
A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys, and serosal membranes.

Cor pulmonale
Right-sided heart failure resulting from chronic hypertension in the pulmonary arteries and right ventricle.

Respiratory tract infection
An infection of the upper or lower respiratory tract.

White papule
A papule with white color.

Emphysema

Reduced forced expiratory volume in one second
An abnormal reduction in the amount of air a person can forcefully expel in one second.

Dyspnea
Difficult or labored breathing. Dyspnea is a subjective feeling only the patient can rate, e.g., on a Borg scale.

Edema
An abnormal accumulation of fluid beneath the skin, or in one or more cavities of the body.

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Failure to thrive

Failure to thrive (FTT) refers to a child whose physical growth is substantially below the norm.

Exocrine pancreatic insufficiency
Impaired function of the exocrine pancreas associated with a reduced ability to digest foods because of lack of digestive enzymes.

Halitosis
Noticeably unpleasant odors exhaled in breathing.

Immunodeficiency
Failure of the immune system to protect the body adequately from infection, due to the absence or insufficiency of some component process or substance.

Hepatosplenomegaly
Simultaneous enlargement of the liver and spleen.

Oligospermia
Reduced count of spermatozoa in the semen, defined as a sperm count below 20 million per milliliter semen.

Clubbing
Broadening of the soft tissues (non-edematous swelling of soft tissues) of the digital tips in all dimensions associated with an increased longitudinal and lateral curvature of the nails.

Azoospermia
Absence of any measurable level of sperm in his semen.

Chest pain
An unpleasant sensation characterized by physical discomfort (such as pricking, throbbing, or aching) localized to the chest.

Leukocytosis
An abnormal increase in the number of leukocytes in the blood.

Reduced forced vital capacity
An abnormal reduction in the amount of air a person can expel following maximal inspiration.

Autosomal dominant inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Abdominal pain
An unpleasant sensation characterized by physical discomfort (such as pricking, throbbing, or aching) and perceived to originate in the abdomen.

Atopic dermatitis
Atopic dermatitis (AD) or atopic eczema is an itchy, inflammatory skin condition with a predilection for the

skin flexures. It is characterized by poorly defined erythema with edema, vesicles, and weeping in the acute stage and skin thickening (lichenification) in the chronic stage.

Pancreatic calcification
The presence of abnormal calcium deposition lesions in the pancreas.

Recurrent Haemophilus influenzae infections
Increased susceptibility to Haemophilus influenzae infections as manifested by recurrent episodes of infection by Haemophilus influenzae.

Biliary cirrhosis
Progressive destruction of the small-to-medium bile ducts of the intrahepatic biliary tree, which leads to progressive cholestasis and often end-stage liver disease.

Pancreatitis
The presence of inflammation in the pancreas.

Phenotypic abnormality
A phenotypic abnormality.

Splanchnic vein thrombosis
The term splanchnic vein thrombosis encompasses Budd-Chiari syndrome (hepatic vein thrombosis), extrahepatic portal vein obstruction (EHPVO), and mesenteric vein thrombosis; the word splanchnic is used to refer to the visceral organs (of the abdominal cavity).

Malabsorption
Impaired ability to absorb one or more nutrients from the intestine.

Rectal prolapse
Protrusion of the rectal mucous membrane through the anus.

Increased circulating gonadotropin level
Overproduction of gonadotropins (FSH, LH) by the anterior pituitary gland.

Elevated sweat chloride
An increased concentration of chloride in the sweat.

Decreased circulating antibody level
An abnormally decreased level of immunoglobulin in blood.

Hypercalciuria

Male infertility

Abnormal thrombosis
Venous or arterial thrombosis (formation of blood clots) of spontaneous nature and which cannot be fully explained by acquired risk (e.g. atherosclerosis).

Recurrent lower respiratory tract infections
An increased susceptibility to lower respiratory tract infections as manifested by a history of recurrent lower respiratory tract infections.

Skin plaque
A plaque is a solid, raised, plateau-like (flat-topped) lesion greater than 1 cm in diameter.

Recurrent bronchopulmonary infections
An increased susceptibility to bronchopulmonary infections as manifested by a history of recurrent bronchopulmonary infections.

Clubbing of fingers
Terminal broadening of the fingers (distal phalanges of the fingers).

Myocardial infarction
Necrosis of the myocardium caused by an obstruction of the blood supply to the heart and often associated with chest pain, shortness of breath, palpitations, and anxiety as well as characteristic EKG findings and elevation of serum markers including creatine kinase-MB fraction and troponin.

Bronchiectasis
Persistent abnormal dilatation of the bronchi owing to localized and irreversible destruction and widening of the large airways.

Decreased testicular size
Reduced volume of the testicle (the male gonad).

Palmoplantar hyperhidrosis
An abnormally increased perspiration on palms and soles.

Recurrent pancreatitis
A recurrent form of pancreatitis.

Fever
Body temperature elevated above the normal range.

Pleural effusion
The presence of an excessive amount of fluid in the pleural cavity.

Cirrhosis
A chronic disorder of the liver in which liver tissue becomes scarred and is partially replaced by regenerative nodules and fibrotic tissue resulting in loss of liver function.

Hepatomegaly
Abnormally increased size of the liver.

Diarrhea
Abnormally increased frequency of loose or watery bowel movements.

Hemoptysis

Coughing up (expectoration) of blood or blood-streaked sputum from the larynx, trachea, bronchi, or lungs.

Steatorrhea
Greater than normal amounts of fat in the feces. This is a result of malabsorption of lipids in the small intestine and results in frothy foul-smelling fecal matter that floats.

Decreased forced expiratory flow 25-75%
A reduction compared to the predicted value of the forced expiratory flow over the middle one-half of the FVC; the average flow from the point at which 25% of the FVC has been exhaled to the point at which 75% of the FVC has been exhaled.

Ileus
Acute obstruction of the intestines preventing passage of the contents of the intestines.

Abnormal enzyme/coenzyme activity
An altered ability of any enzyme or their cofactors to act as catalysts. This term includes changes due to altered levels of an enzyme.

Meconium ileus
Obstruction of the intestine due to abnormally thick meconium.

Abnormal renal morphology
Any structural anomaly of the kidney.

Palmoplantar keratoderma
Abnormal thickening of the skin of the palms of the hands and the soles of the feet.

Chronic sinusitis
A chronic form of sinusitis.

Palmar pruritus
Pruritus is an itch or a sensation that makes a person want to scratch. This term refers to an abnormally increased sensation of itching over the palm(s) of the hand.

Crackles
Crackles are discontinuous, explosive, and nonmusical adventitious lung sounds normally heard in inspiration and sometimes during expiration. Crackles are usually classified as fine and coarse crackles based on their duration, loudness, pitch, timing in the respiratory cycle, and relationship to coughing and changing body position.

Heterogeneous

Non-obstructive azoospermia
Absence of any measurable level of sperm in his semen, resulting from a defect in the production of spermatozoa in the testes. This can be differentiated from obstructive azoospermia on the basis of testicular biopsy.

Abnormal phalangeal joint morphology of the hand

Decreased pulmonary function

Abnormal spermatogenesis
Incomplete maturation or aberrant formation of the male gametes.

Diabetes mellitus
A group of abnormalities characterized by hyperglycemia and glucose intolerance.

Cachexia
Severe weight loss, wasting of muscle, loss of appetite, and general debility related to a chronic disease.

Wheezing
A high-pitched whistling sound associated with labored breathing.

Dehydration

Recurrent pneumonia
An increased susceptibility to pneumonia as manifested by a history of recurrent episodes of pneumonia.

Asthma
Asthma is characterized by increased responsiveness of the tracheobronchial tree to multiple stimuli, leading to narrowing of the air passages with resultant dyspnea, cough, and wheezing.

Excessive skin wrinkling on dorsum of hands and fingers

Recurrent sinopulmonary infections
An increased susceptibility to infections involving both the paranasal sinuses and the lungs, as manifested by a history of recurrent sinopulmonary infections.

Productive cough
A cough that produces phlegm or mucus.