

## Gene

**TP53**

## Associated Diseases

Pancreatic Cancer  
Osteosarcoma  
Li-fraumeni Syndrome  
Essential Thrombocythemia  
Colorectal Cancer  
Osteosarcoma  
Hepatocellular Carcinoma  
Adrenocortical Carcinoma  
Papilloma Of Choroid Plexus  
Li-fraumeni Syndrome  
Nasopharyngeal Carcinoma  
Papilloma Of Choroid Plexus  
Bone Marrow Failure Syndrome 5  
Glioma Susceptibility 1  
Adrenocortical Carcinoma, Pediatric  
Familial Pancreatic Carcinoma  
Hereditary Breast And Ovarian Cancer Syndrome  
Breast Cancer

## Phenotype

### **Jaundice**

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

### **Panic attack**

A sudden episode of intense fear in a situation in which there is no danger or apparent cause.

### **Abnormal cerebral vascular morphology**

An anomaly of the cerebral blood vessels.

### **Hereditary nonpolyposis colorectal carcinoma**

### **Poor appetite**

A reduced desire to eat.

### **Acute myeloid leukemia**

A form of leukemia characterized by overproduction of an early myeloid cell.

### **Neoplasm of the pancreas**

A tumor (abnormal growth of tissue) of the pancreas.

**Microcephaly**

Head circumference below 2 standard deviations below the mean for age and gender.

**Weight loss**

Reduction of total body weight.

**Nausea and vomiting**

Nausea is a commonly encountered symptom that has been defined as an unpleasant painless subjective feeling that one will imminently vomit. Vomiting has been defined as the forceful expulsion of the contents of the stomach, duodenum, or jejunum through the oral cavity. While nausea and vomiting are often thought to exist on a temporal continuum, this is not always the case. There are situations when severe nausea may be present without emesis and less frequently, when emesis may be present without preceding nausea.

**Ependymoma**

The presence of an ependymoma of the central nervous system.

**Nausea**

A sensation of unease in the stomach together with an urge to vomit.

**Breast carcinoma**

The presence of a carcinoma of the breast.

**Intermittent diarrhea**

Repeated episodes of diarrhea separated by periods without diarrhea.

**Increased circulating lactate dehydrogenase concentration**

An elevated level of the enzyme lactate dehydrogenase in the blood circulation.

**Hepatosplenomegaly**

Simultaneous enlargement of the liver and spleen.

**Melanoma**

The presence of a melanoma, a malignant cancer originating from pigment producing melanocytes. Melanoma can originate from the skin or the pigmented layers of the eye (the uvea).

**Arterial thrombosis**

The formation of a blood clot inside an artery.

**Abnormal circulating dehydroepiandrosterone concentration**

A deviation from the normal concentration of dehydroepiandrosterone in the circulation.

**Hypokalemia**

An abnormally decreased potassium concentration in the blood.

**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.

### **Headache**

Cephalgia, or pain sensed in various parts of the head, not confined to the area of distribution of any nerve.

### **Splenomegaly**

Abnormal increased size of the spleen.

### **Leukemia**

A cancer of the blood and bone marrow characterized by an abnormal proliferation of leukocytes.

### **Abnormality of metabolism/homeostasis**

### **Abnormality of the metaphysis**

An abnormality of one or more metaphysis, i.e., of the somewhat wider portion of a long bone that is adjacent to the epiphyseal growth plate and grows during childhood.

### **Abnormality of reproductive system physiology**

An abnormal functionality of the genital system.

### **Colon cancer**

### **Neoplasm of the larynx**

### **Thyroid carcinoma**

The presence of a carcinoma of the thyroid gland.

### **Hypertension**

The presence of chronic increased pressure in the systemic arterial system.

### **Lymphadenopathy**

Enlargement (swelling) of a lymph node.

### **Hypogonadism**

A decreased functionality of the gonad.

### **Cognitive impairment**

Abnormal cognition with deficits in thinking, reasoning, or remembering.

### **Acute lymphoblastic leukemia**

A form of acute leukemia characterized by excess lymphoblasts.

### **Neoplasm of the rectum**

### **Palpitations**

A sensation that the heart is pounding or racing, which is a non-specific sign but may be a manifestation of arrhythmia.

### **Vomiting**

Forceful ejection of the contents of the stomach through the mouth by means of a series of involuntary spasmodic contractions.

### **Anorexia**

A lack or loss of appetite for food (as a medical condition).

### **Delayed skeletal maturation**

A decreased rate of skeletal maturation. Delayed skeletal maturation can be diagnosed on the basis of an estimation of the bone age from radiographs of specific bones in the human body.

### **Increased megakaryocyte count**

Increased megakaryocyte number, i.e., of platelet precursor cells, present in the bone marrow.

### **Adrenocorticotrophic hormone deficiency**

A reduced ability to secrete adrenocorticotrophic hormone (ACTH), a hormone that stimulates the adrenal cortex to secrete of glucocorticoids such as cortisol.

### **Abnormality of the femoral metaphysis**

An anomaly of the femoral metaphysis.

### **Anemia**

A reduction in erythrocytes volume or hemoglobin concentration.

### **Paresthesia**

Abnormal sensations such as tingling, pricking, or numbness of the skin with no apparent physical cause.

### **Increased serum estradiol**

An elevation above normal limits of the concentration of estradiol in the circulation.

### **Rhabdomyosarcoma**

### **Pure red cell aplasia**

A type of anemia resulting from suppression of erythropoiesis with little or no abnormality of leukocyte or platelet production. Erythroblasts are virtually absent in bone marrow; however, leukocyte and platelet production show little or no reduction.

### **Soft tissue sarcoma**

A type of sarcoma (A connective tissue neoplasm formed by proliferation of mesodermal cells) that develops from soft tissues like fat, muscle, nerves, fibrous tissues, blood vessels, or deep skin tissues.

**Fever**

Body temperature elevated above the normal range.

**Abnormality of the tibial metaphysis****Increased circulating androstenedione concentration**

Increased concentration of androstenedione in the blood circulation.

**Muscle weakness**

Reduced strength of muscles.

**Somatic mutation**

A mode of inheritance in which a trait or disorder results from a de novo mutation occurring after conception, rather than being inherited from a preceding generation.

**Subacute progressive viral hepatitis****Stomach cancer**

A cancer arising in any part of the stomach.

**Increased level of L-fucose in urine**

An increase in the level of L-fucose in the urine.

**Neoplasm of the liver**

A tumor (abnormal growth of tissue) of the liver.

**Hodgkin lymphoma**

A type of lymphoma characterized microscopically by multinucleated Reed-Sternberg cells.

**Renal cell carcinoma**

A type of carcinoma of the kidney with origin in the epithelium of the proximal convoluted renal tubule.

**Papilloma**

A tumor of the skin or mucous membrane with finger-like projections.

**Neoplasm of the central nervous system**

A neoplasm of the central nervous system.

**Hypertonia**

A condition in which there is increased muscle tone so that arms or legs, for example, are stiff and difficult to move.

**Central primitive neuroectodermal tumor**

A primitive neuroectodermal neoplasm that occurs in the central nervous system.

**Prostate cancer**

A cancer of the prostate.

**Neoplasm of the lung**

Tumor of the lung.

**Colorectal polyposis**

Multiple abnormal growths that arise from the lining of the large intestine (colon or rectum) and protrude into the intestinal lumen.

**Non-Hodgkin lymphoma**

A type of lymphoma characterized microscopically by the absence of multinucleated Reed-Sternberg cells.

**Choroid plexus carcinoma**

Intraventricular papillary neoplasm derived from choroid plexus epithelium. Plexus tumors are most common in the lateral and fourth ventricles; while 80% of lateral ventricle tumors present in children, fourth ventricle tumors are evenly distributed in all age groups. Clinically, choroid plexus tumors tend to cause hydrocephalus and increased intracranial pressure. Histologically, choroid plexus papillomas correspond to WHO grade I, choroid plexus carcinomas to WHO grade III.

**Seizure**

A seizure is an intermittent abnormality of nervous system physiology characterised by a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain.

**Polygenic inheritance**

A type of multifactorial inheritance governed by the simultaneous action of many (more than three) gene loci.

**Abnormal platelet morphology**

An anomaly in platelet form, ultrastructure, or intracellular organelles.

**Osteosarcoma**

A malignant bone tumor that usually develops during adolescence and usually affects the long bones including the tibia, femur, and humerus. The typical symptoms of osteosarcoma comprise bone pain, fracture, limitation of motion, and tenderness or swelling at the site of the tumor.

**Transitional cell carcinoma of the bladder**

The presence of a carcinoma of the urinary bladder with origin in a transitional epithelial cell.

**Hemiplegia/hemiparesis**

Loss of strength in the arm, leg, and sometimes face on one side of the body. Hemiplegia refers to a severe or complete loss of strength, whereas hemiparesis refers to a relatively mild loss of strength.

**Chronic fatigue**

Subjective feeling of tiredness characterized by a lack of energy and motivation that persists for six months or longer.

**Hyperaldosteronism**

Overproduction of the mineralocorticoid aldosterone by the adrenal cortex.

**Elevated hepatic transaminase**

Elevations of the levels of SGOT and SGPT in the serum. SGOT (serum glutamic oxaloacetic transaminase) and SGPT (serum glutamic pyruvic transaminase) are transaminases primarily found in the liver and heart and

are released into the bloodstream as the result of liver or heart damage. SGOT and SGPT are used clinically mainly as markers of liver damage.

**Amaurosis fugax**

A transient visual disturbance that is typically caused by a circulatory, ocular or neurological underlying condition.

**Medulloblastoma**

A rapidly growing embryonic tumor arising in the posterior part of the cerebellar vermis and neuroepithelial roof of the fourth ventricle in children. More rarely, medulloblastoma arises in the cerebellum in adults.

**Prolonged bleeding time**

Prolongation of the time taken for a standardized skin cut of fixed depth and length to stop bleeding.

**Increased urinary cortisol level**

Abnormally increased concentration of cortisol in the urine.

**Hyperhidrosis**

Abnormal excessive perspiration (sweating) despite the lack of appropriate stimuli like hot and humid weather.

**Intestinal pseudo-obstruction**

A functional rather than mechanical obstruction of the intestines, associated with manifestations that resemble those caused by an intestinal obstruction, including distension, abdominal pain, nausea, vomiting, constipation or diarrhea, in an individual in whom a mechanical blockage has been excluded.

**Neoplasia of the nasopharynx****Anxiety**

Intense feelings of nervousness, tenseness, or panic, often in reaction to interpersonal stresses; worry about the negative effects of past unpleasant experiences and future negative possibilities; feeling fearful, apprehensive, or threatened by uncertainty; fears of falling apart or losing control.

**Adrenocortical carcinoma**

A malignant neoplasm of the adrenal cortex that may produce hormones such as cortisol, aldosterone, estrogen, or testosterone.

**Exocrine pancreatic insufficiency**

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

**Neoplasm of the gastrointestinal tract**

A tumor (abnormal growth of tissue) of the gastrointestinal tract.

**Myelodysplasia**

Clonal hematopoietic stem cell disorders characterized by dysplasia (ineffective production) in one or more hematopoietic cell lineages, leading to anemia and cytopenia.

**Elevated circulating alkaline phosphatase concentration**

Abnormally increased serum levels of alkaline phosphatase activity.

**Hepatocellular carcinoma**

A kind of neoplasm of the liver that originates in hepatocytes and presents macroscopically as a soft and hemorrhagic tan mass in the liver.

**Short stature**

A height below that which is expected according to age and gender norms. Although there is no universally accepted definition of short stature, many refer to "short stature" as height more than 2 standard deviations below the mean for age and gender (or below the 3rd percentile for age and gender dependent norms).

**Chest pain**

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

**Nephroblastoma**

The presence of a nephroblastoma, which is a neoplasm of the kidney that primarily affects children.

**Elevated serum 11-deoxycortisol**

Increased concentration of 11-deoxycortisol in the circulation. 11-deoxycorticosterone, which is also known as simply deoxycorticosterone and 21-hydroxyprogesterone, is a steroid hormone that is produced in the adrenals and is a precursor to aldosterone.

**Abnormal lactate dehydrogenase level**

A deviation from the normal serum concentration/activity of lactate dehydrogenase (LDH), which catalyzes the reduction of pyruvate to form lactate.

**Acute leukemia**

A clonal (malignant) hematopoietic disorder with an acute onset, affecting the bone marrow and the peripheral blood. The malignant cells show minimal differentiation and are called blasts, either myeloid blasts (myeloblasts) or lymphoid blasts (lymphoblasts).

**Joint swelling****Increased circulating androgen concentration**

An elevation of the blood concentration of an androgen, that is, of a steroid hormone that controls development and maintenance of masculine characteristics. The androgens include testosterone and Dehydroepiandrosterone.

**Increased body weight**

Abnormally increased body weight.

**Hypertrichosis**

Hypertrichosis is increased hair growth that is abnormal in quantity or location.

**Transient ischemic attack**



### **Choriocarcinoma**

A malignant, trophoblastic and aggressive cancer, usually of the placenta. It is characterized by early hematogenous spread to the lungs and belongs to the far end of the spectrum of gestational trophoblastic disease (GTD), a subset of germ cell tumors.

### **Extrahepatic cholestasis**

Impairment of bile flow due to obstruction in large bile ducts outside the liver.

### **Striae distensae**

Thinned, erythematous, depressed bands of atrophic skin. Initially, striae appear as flattened and thinned, pinkish linear regions of the skin. Striae tend to enlarge in length and become reddish or purplish. Later, striae tend to appear as white, depressed bands that are parallel to the lines of skin tension. Striae distensae occur most often in areas that have been subject to distension such as the lower back, buttocks, thighs, breast, abdomen, and shoulders.

### **Irritability**

A proneness to anger, i.e., a condition of being easily bothered or annoyed.

### **Hydrocephalus**

Hydrocephalus is an active distension of the ventricular system of the brain resulting from inadequate passage of CSF from its point of production within the cerebral ventricles to its point of absorption into the systemic circulation.

### **Pancreatic adenocarcinoma**

The presence of an adenocarcinoma of the pancreas.

### **Abnormality of urine homeostasis**

An abnormality of the composition of urine or the levels of its components.

### **Prostate neoplasm**

### **Uterine leiomyosarcoma**

The presence of a leiomyosarcoma of the uterus.

### **Choroid plexus papilloma**

Choroid plexus papilloma is a histologically benign neoplasm located in the ventricular system of the choroid plexus.

### **Neoplasm**

An organ or organ-system abnormality that consists of uncontrolled autonomous cell-proliferation which can occur in any part of the body as a benign or malignant neoplasm (tumour).

### **Decreased circulating antibody level**

An abnormally decreased level of immunoglobulin in blood.

### **Abnormal thrombocyte morphology**

An abnormality of platelets.

**Neoplasm of the stomach**

A tumor (abnormal growth of tissue) of the stomach.

**Myelofibrosis**

Replacement of bone marrow by fibrous tissue.

**Glioblastoma multiforme**

A tumor arising from glia in the central nervous system with macroscopic regions of necrosis and hemorrhage. Microscopically, glioblastoma multiforme is characterized by regions of pseudopalisading necrosis, pleomorphic nuclei and cells, and microvascular proliferation.

**Abnormal nervous system morphology**

A structural anomaly of the nervous system.

**Primary peritoneal carcinoma**

A type of cancer that originates in the peritoneum. It is to be distinguished from metastatic cancer of the peritoneum. Peritoneal cancer can occur anywhere in the abdominal space, and affects the surface of organs contained inside the peritoneum.

**Back pain**

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

**Abnormality of bone marrow cell morphology**

An anomaly of the form or number of cells in the bone marrow.

**Renal neoplasm**

The presence of a neoplasm of the kidney.

**Papilledema**

Papilledema refers to edema (swelling) of the optic disc secondary to any factor which increases cerebral spinal fluid pressure.

**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.

**Global developmental delay**

A delay in the achievement of motor or mental milestones in the domains of development of a child, including motor skills, speech and language, cognitive skills, and social and emotional skills. This term should only be used to describe children younger than five years of age.

**Testicular atrophy**

Wasting (atrophy) of the testicle (the male gonad) manifested by a decrease in size and potentially by a loss of fertility.

**Lung adenocarcinoma**

**Astrocytoma**

Astrocytoma is a neoplasm of the central nervous system derived from astrocytes. Astrocytes are a type of glial cell, and thus astrocytoma is a subtype of glioma.

**Pain**

An unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage.

**Testicular neoplasm**

The presence of a neoplasm of the testis.

**Osteolysis**

Osteolysis refers to the destruction of bone through bone resorption with removal or loss of calcium.

**Visual impairment**

Visual impairment (or vision impairment) is vision loss (of a person) to such a degree as to qualify as an additional support need through a significant limitation of visual capability resulting from either disease, trauma, or congenital or degenerative conditions that cannot be corrected by conventional means, such as refractive correction, medication, or surgery.

**Paradoxical increased cortisol secretion on dexamethasone suppression test****Venous thrombosis**

Formation of a blood clot (thrombus) inside a vein, causing the obstruction of blood flow.

**Retinoblastoma**

A tumor of the eye originating from cells of the retina.

**Lymphoma**

A cancer originating in lymphocytes and presenting as a solid tumor of lymphoid cells.

**Functional intestinal obstruction****Ovarian neoplasm**

A tumor (abnormal growth of tissue) of the ovary.

**Heterogeneous****Pathologic fracture**

A pathologic fracture occurs when a bone breaks in an area that is weakened secondarily to another disease process such as tumor, infection, and certain inherited bone disorders. A pathologic fracture can occur without a degree of trauma required to cause fracture in healthy bone.

**Abnormal fallopian tube morphology**

An abnormality of the fallopian tube.

**Diabetes mellitus**

A group of abnormalities characterized by hyperglycemia and glucose intolerance.

**Micronodular cirrhosis**

A type of cirrhosis characterized by the presence of small regenerative nodules.

**Ovarian carcinoma**

A malignant neoplasm originating from the surface ovarian epithelium.

**Neoplasm of head and neck**

A tumor (abnormal growth of tissue) of the head and neck region with origin in the lip, oral cavity, nasal cavity, paranasal sinuses, pharynx, or larynx.

**Peritoneal abscess**

The presence of an abscess of the peritoneum.

**Increased circulating cortisol level**

Overproduction of the hormone of cortisol by the adrenal cortex, resulting in a characteristic combination of clinical symptoms termed Cushing syndrome, with truncal obesity, a round, full face, striae atrophicae and acne, muscle weakness, and other features.