

Gene

CHEK2

Associated Diseases

Hereditary Breast And Ovarian Cancer Syndrome
Colorectal Cancer
Prostate Cancer
Li-fraumeni Syndrome
Breast Cancer
Osteosarcoma
Li-fraumeni Syndrome 2
Osteosarcoma

Phenotype

Colorectal polyposis

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

Hereditary nonpolyposis colorectal carcinoma

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.

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.

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.

Glioma

The presence of a glioma, which is a neoplasm of the central nervous system originating from a glial cell (astrocytes or oligodendrocytes).

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.

Weight loss

Reduction of total body weight.

Ependymoma

The presence of an ependymoma of the central nervous system.

Adrenocortical carcinoma

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

Breast carcinoma

The presence of a carcinoma of the breast.

Neoplasm of the gastrointestinal tract

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

Increased circulating lactate dehydrogenase concentration

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

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.

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).

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.

Joint swelling**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.

Leukemia

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

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.

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.

Colon cancer**Neoplasm of the larynx****Thyroid carcinoma**

The presence of a carcinoma of the thyroid gland.

Uterine leiomyosarcoma

The presence of a leiomyosarcoma of the uterus.

Acute lymphoblastic leukemia

A form of acute leukemia characterized by excess lymphoblasts.

Neoplasm of the rectum**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).

Neoplasm of the stomach

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

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.

Abnormality of the femoral metaphysis

An anomaly of the femoral metaphysis.

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.

Renal neoplasm

The presence of a neoplasm of the kidney.

Rhabdomyosarcoma**Fever**

Body temperature elevated above the normal range.

Abnormality of the tibial metaphysis**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.

Meningioma

The presence of a meningioma, i.e., a benign tumor originating from the dura mater or arachnoid mater.

Testicular neoplasm

The presence of a neoplasm of the testis.

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.

Osteolysis

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

Retinoblastoma

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

Stomach cancer

A cancer arising in any part of the stomach.

Lymphoma

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

Ovarian neoplasm

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

Sarcoma

A connective tissue neoplasm formed by proliferation of mesodermal cells. Bone and soft tissue sarcomas are the main types of sarcoma. Sarcoma is usually highly malignant.

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.

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.

Abnormal fallopian tube morphology

An abnormality of the fallopian tube.

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.

Neoplasm of the central nervous system

A neoplasm of the central nervous system.

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.